SYNLAB MVZ Humangenetik Mannheim

Stand: 07.01.2026

Whole Exome Sequencing (WES)

Whole Exome Sequencing (WES) : Datenanalyse auf Grundlage des Exoms inklusive mitochondrialem Genom. Hierzu wird die Angabe der klinischen Symptomatik des Patienten (HPO-Terms) benötigt.

Die WES-Analyse sollte vorzugsweise als Trio-WES (Eltern und Indexpatient) durchgeführt werden, kann aber auch mit eingeschränkter Aussagekraft als Solo-Exom (nur Indexpatient) durchgeführt werden.

Weitere Informationen finden Sie unter **www.zhma.de.**

Next Generation Sequencing (NGS)

Die in **Kapitel 11.4.2 des EBM** gelisteten indikationsbezogenen Untersuchungen enthalten eine eindeutige diagnostische Vorgehensweise. Eine diagnostische Anforderung und Durchführung der Analyse muss exakt wie im Kapitel 11.4.2 beschrieben **(Stufendiagnostik)** erfolgen.
Die Diagnostik in Kapitel 11.4.2 ist abschließend, d. h. unter ein und derselben Verdachtsdiagnose bzw. Indikation ist im Krankheitsfall (d. h. innerhalb von 1 Jahr) keine weitere Diagnostik möglich.

Die Genehmigungspflicht für die erweiterte Mutationssuche über 25 kb (Streichung der GOP 11514) wurde aufgehoben.
Ab sofort können somit bei entsprechender Indikation Multigen-Panels, deren Größe 25 kb überschreitet, ohne vorherigen Antrag auf Kostenübernahme bei der gesetzlichen Krankenversicherung beauftragt werden. Für privatversicherte Patienten sowie private Kostenträger (Krankenhäuser etc.) können auf Wunsch entsprechende Kostenvoranschläge erstellt werden.

Hinweis: Vor Durchführung einer Leistung nach den Gebührenordnungspositionen der Abschnitte 11.4.2 und 11.4.3 sind die Voraussetzungen gemäß § 6 der Qualitätssicherungsvereinbarung Molekulargenetik gemäß § 135 Abs. 2 SGB V zu überprüfen und, falls erforderlich, festzustellen.

Weitere Informationen finden Sie unter **www.zhma.de/einsenderinformationen** . **Humangenetische Leistungen sind NICHT budget-relevant.**

Untersuchungsmaterial

Untersuchungsmaterial:
3 - 5 ml EDTA-Blut (E), Fruchtwasser (FW), Chorionzotten (C), Abortgewebe (A), Tumorgewebe (T),
2 - 3 Wangenschleimhautabstriche (WA), Fibroblastenkultur (F) des betroffenen Gewebes

Ein aktuelles Gesamtverzeichnis unserer Leistungen und aktuelle Einsenderinformationen finden Sie unter **www.zhma.de**

Bitte beachten Sie auch unser **"Handbuch der Primärprobengewinnung".**

Panel-Verzeichnis

Krankheitskategorien

Whole Exome Sequencing (WES)

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Achromatopsie (ACHM) * Gen-Panel: ID164.02, 6 Gene (10,4 kb) ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	3 - 5 Wo	E
Albinismus, umfassende Diagnostik * Gen-Panel: ID175.05 Albinismus, umfassende Diagnostik: 33 Gene (73,8 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1 Okulokutaner Albinismus (OCA, OA): 9 Gene (13,2 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 Waardenburg-Syndrom (WS): 8 Gene (9,3 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Griscelli-Syndrom (GS): 3 Gene (8,0 kb) MLPH, MYO5A, RAB27A	3 - 5 Wo	E
Altersbedingte Makuladegeneration (AMD, ARMD) * Gen-Panel: ID186.00, 16 Gene (51,0 kb) ABCA4, APOE, ARMS2, C2, C3, C9, CFB, CFH, CFI, CST3, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, RAX2	3 - 5 Wo	E
Anophthalmie und Mikrophthalmie (MCOP) * Gen-Panel ID263.02 Anophthalmie und Mikrophthalmie (MCOP): 46 Gene (107,6 kb) ABCB6, ALDH1A3, BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3, FZD5, GDF3, GDF6, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, MITF, NAA10, NHEJ1, OTX2, PAX2, PAX6, PITX3, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, TMEM98, VAX1, VSX2, YAP1 Mikrophthalmie, isolierte Form (MCOP): 6 Gene (8,6 kb) ALDH1A3, GDF3, GDF6, MFRP, PRSS56, VSX2 Mikrophthalmie, syndromale Form (MCOPS): 13 Gene (24,9 kb) BCOR, BMP4, HCCS, HMGB3, MAB21L2, NAA10, OTX2, SOX2, RARB, RAX, STRA6, TENM3, VAX1 Mikrophthalmie und Kolobom (MCOPCB): 11 Gene (22,1 kb) ABCB6, GDF3, GDF6, FZD5, NHEJ1, PAX6, RBP4, SHH, STRA6, TENM3, VSX2 Syndrome mit Mikrophthalmie: 34 Gene (90,4 kb) BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MITF, NAA10, OTX2, PAX2, PITX3, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TFAP2A, TMEM98, TENM3, VAX1, YAP1	4 - 6 Wo	E
Dysgenesie des vorderen Augensegmentes (ASGD) * Gen-Panel: ID182.03 Dysgenesie des vorderen Augensegmentes (ASGD): 11 Gene (21,5 kb) B3GLCT, CPAMD8, CYP1B1, ELP4, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN, TRIM44 Peters-Anomalie: 7 Gene (8,8 kb) B3GLCT, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3 Axenfeld-Rieger-Anomalie: 3 Gene (3,8 kb) FOXC1, PAX6, PITX2 Aniridie (AN): 3 Gene (3,6 kb) ELP4, PAX6, TRIM44	3 - 5 Wo	E
Fraser-Syndrom (FRASRS) * Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
Fuchs-Endotheldystrophie (FECD) * Gen-Panel: ID261.00, 5 Gene (13,5 kb) AGBL1, COL8A2, SLC4A11, TCF4, ZEB1	3 - 5 Wo	E
Glaukom (GLC) * Gen-Panel: ID275.01 Glaukom (GLC): 27 Gene (53,5 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CPAMD8, CYP1B1, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, PAX6, PITX2, PITX3, PXDN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMCO1, WDR36 Primäres Glaukom (GLC1, GLC3): 20 Gene (37,8 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CYP1B1, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMCO1, WDR36 Dysgenesie des vorderen Augensegmentes (ASGD): 8 Gene (17,5 kb) CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS) * Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
Hohe Myopie (MYP) * Gen-Panel ID079.03 Hohe Myopie (MYP): 25 Gene (81,1 kb) ARR3, CACNA1F, CNGB3, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CPSF1, GPR179, GRM6, GZF1, IRX5, LOXL3, LRPAP1, LRP2, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644 Hohe Myopie, nicht-syndromale Form (MYP): 12 Gene (24,5 kb) ARR3, CNGB3, CPSF1, LOXL3, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644 Syndrome mit hoher Myopie: 14 Gene (58,7 kb) CACNA1F, COL11A1, COL2A1, COL9A1, COL9A2, COL9A3, COL18A1, GPR179, GRM6, GZF1, LRP2, IRX5, P3H2, SLITRK6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hornhautdystrophie *		
Gen-Panel: ID329.01		
Hornhautdystrophie: 27 Gene (68,9 kb)		
AGBL1, CHST6, COL8A2, COL17A1, CYP4V2, DCN, GRHL2, GSN, KERA, KRT3, KRT12, LCAT, LOXHD1, MCOLN1, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469		
Fuchs-Endotheldystrophie (FECD): 6 Gene (20,2 kb)		
AGBL1, COL8A2, LOXHD1, SLC4A11, TCF4, ZEB1		
Hintere polymorphe Hornhautdystrophie (PPCD): 4 Gene (8,2 kb)		
COL8A2, GRHL2, OVOL2, ZEB1		
Gittrige Hornhautdystrophie (CDL): 3 Gene (5,4 kb)	3 - 5 Wo	E
GSN, TACSTD2, TGFBI		
Stromale Hornhautdystrophie: 5 Gene (11,6 kb)		
CHST6, DCN, PIKFYVE, TGFBI, UBIAD1		
Epitheliale Hornhautdystrophie: 6 Gene (12,6 kb)		
COL17A1, KRT3, KRT12, MCOLN1, TACSTD2, TGFBI		
Brittle-Cornea-Syndrom (BCN): 2 Gene (13,8 kb)		
PRDM5, ZNF468		
Keratokonus (KTCN): 2 Gene (2,5 kb)		
TUBA3D, VSX1		
Katarakt (CTRCT) *		
Gen-Panel: ID206.01		
Katarakt (CTRCT): 37 Gene (59,7 kb)		
AGK, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, DNMBP, EPHA2, FOXE3, FYCO1, GCNT2, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, NHS, PITX3, SIPA1L3, SLC16A12, TDRD7, UNC45B, VIM, WFS1		
Katarakt (CTRCT), autosomal-dominant: 26 Gene (29,2 kb)	3 - 5 Wo	E
BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, EPHA2, GJA3, GJA8, HSF4, MAF, MIP, PITX3, SLC16A12, UNC45B, VIM, WFS1		
Katarakt (CTRCT), autosomal-rezessiv: 15 Gene (30,0 kb)		
AGK, BFSP1, CRYAA, CRYAB, CRYBB1, CRYBB3, DNMBP, FOXE3, FYCO1, GCNT2, LEMD2, LIM2, LSS, SIPA1L3, TDRD7		
Katarakt (CTRCT), X-chromosomal: 1 Gene (4,9 kb)		
NHS		
Kongenitale extraokuläre Muskelfibrose (CFEOM) *		
Gen-Panel: ID063.00, 5 Gene (10,5 kb)	3 - 5 Wo	E
COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3		
Kongenitaler Nystagmus (NYS) *		
Gen-Panel ID331.01		
Kongenitaler Nystagmus (NYS): 31 Gene (71,0 kb)		
AHR, ATF6, CACNA1F, CEP290, CNGA3, CNGB3, CRB1, CRX, DAGLA, DCT, FRMD7, GPR143, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, LRMDA, NMNAT1, NYX, OCA2, PAX6, ROBO1, RPE65, RPGRIP1, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1		
Nystagmus, nicht-syndromal (NYS): 3 Gene (8,3 kb)		
FRMD7, GPR143, ROBO1	3 - 5 Wo	E
Foveahypoplasie, nicht-syndromal (FVH): 3 Gene (5,1 kb)		
AHR, PAX6, SLC38A8		
Okulokutaner Albinismus (OCA): 7 Gene (11,1 kb)		
DCT, LRMDA, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Lebersche kongenitale Amaurose (LCA): 12 Gene (29,5 kb)		
CEP290, CRB1, CRX, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1		
Kongenitale stationäre Nachtblindheit (CSNB) *		
Gen-Panel: ID267.00, 14 Gene (39,2 kb)	3 - 5 Wo	E
CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, SAG, RHO, SLC24A1, TRPM1		
Lebersche hereditäre Optikusneuropathie (LHON) [CO]		
Gen-Panel: ID701.00, 17 Gene (10,5 kb)	3 - 5 Wo	E
MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT-TQ, MT-TT		
Lebersche kongenitale Amaurose (LCA) *		
Gen-Panel: ID187.01, 19 Gene (38,8 kb)	3 - 5 Wo	E
AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45		
Netzhauterkrankungen, umfassende Diagnostik *		
Gen-Panel: ID383.01, 302 Gene (761,8 kb)		
ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARSG, ATF6, ATOH7, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP78, CERKL, CFAP20, CFAP410, CFAP418, CFH, CHM, CIB2, CLCC1, CLEC3B, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COQ2, COQ8B, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTNND1, CTSD, CWC27, CYP4V2, DCT, DHDDS, DHX38, DNAJC5, DRAM2, DYNC2H1, EFEMP1, ELOVL4, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FSCN2, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HGSNAT, HK1, HKDC1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF3B, KIF11, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MAN2B1, MAPKAPK3, MCOLN1, MED12, MERTK, MFRP, MFSD8, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTTP, MVK, MYO7A, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDZD7, PDE6H, PDSS1, PEX1, PEX2, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, POMGNT2, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PYGM, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS2, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SAMD7, SCAPER, SDCCAG8, SEMA4A, SGSH, SLC24A1, SLC38A8, SLC6A6, SLC7A14, SNRNP200, SPATA7, SRD5A3, SSBP1, STN1, STX3, SUMF1, TIMM8A, TIMP3, TINF2, TLCD3B, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, VWA8, WDPCP, WDR19, WHRN, ZFYVE26, ZNF408, ZNF423, ZNF513	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Makuladystrophie (MD) *	3 - 5 Wo	E
Gen-Panel: ID139.03, 22 Gene (49,6 kb)		
ABCA4, BEST1, CDH3, CDHR1, CHST6, CLEC3B, CNGB3, CRB1, CRX, CTNNA1, ELOVL4, IMPG1, IMPG2, MAPKAPK3, MFSD8, PROM1, PRPH2, RDH8, RDH12, RP1L1, SIX6, TIMP3		
Morbus Stargardt (STGD) *	3 - 5 Wo	E
Gen-Panel: ID102.01, 4 Gene (11,4 kb)		
ABCA4, ELOVL4, PROM1, PRPH2		
Okulokutaner Albinismus (OCA) *	3 - 5 Wo	E
Gen-Panel: ID082.02, 9 Gene (13,3 kb)		
DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Optikusatrophie (OPA) *	3 - 5 Wo	E
Gen-Panel ID081.05		
Optikusatrophie (OPA): 38 Gene (60,6 kb)		
ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, KLC2, MCAT, MECR, MFF, MFN2, MIEF1, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, RTN4IP1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, SSBP1, TBCE, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1, ZNHIT3		
Optikusatrophie (OPA) nicht-syndromal: 12 Gene (18,5 kb)		
ACO2, AFG3L2, DNM1L, MCAT, MECR, MIEF1, OPA1, OPA3, RTN4IP1, SSBP1, TMEM126A, YME1L1		
Syndrome mit Optikusatrophie: 29 Gene (46,6 kb)		
ATP1A3, C19ORF12, CISD2, DNAJC30, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, TBCE, TIMM8A, UCHL1, WFS1, ZNHIT3		
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB) *	3 - 5 Wo	E
Gen-Panel: ID300.00		
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB): 10 Gene (17,9 kb)		
DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK		
Progressive externe Ophthalmoplegie, autosomal-dominant (PEOA): 6 Gene (12,4 kb)		
DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK		
Progressive externe Ophthalmoplegie, autosomal-rezessiv (PEOB): 5 Gene (9,2 kb)		
DGUOK, POLG, RNASEH1, TK2, TOP3A		
Retinitis pigmentosa (RP), autosomal-dominant *	3 - 5 Wo	E
Gen-Panel: ID053.03, 29 Gene (64,9 kb)		
AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPG1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS, VWA8		
Retinitis pigmentosa (RP), autosomal-rezessiv *	4 - 6 Wo	E
Gen-Panel: ID050.04, 61 Gene (162,4 kb)		
ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513		
Retinitis pigmentosa (RP), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID288.02		
Retinintis pigmentosa (RP), umfassende Diagnostik: 87 Gene (216,9 kb)		
ABCA4, AGBL5, AHR, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS2, BEST1, CC2D2A, CDHR1, CERKL, CFAP418, CHM, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, DHX38, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, HGSNAT, HK1, HKDC1, IDH3A, IDH3B, IFT140, IFT172, IFT43, IMPDH1, IMPG1, IMPG2, KIAA1549, KIF3B, KIZ, KLHL7, LRAT, MAK, MERTK, NEK2, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF408, ZNF513		
Retinitis pigmentosa (RP), autosomal-dominant: 28 Gene (59,2 kb)		
AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPDH1, IMPG1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS		
Retinitis pigmentosa (RP), autosomal-rezessiv: 61 Gene (162,4 kb)		
ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513		
Retinitis pigmentosa (RP), X-chromosomal: 4 Gene (8,5 kb)		
CHM, RP2, RPGR, OFD1		
Senior-Loken-Syndrom (SLSN) *	3 - 5 Wo	E
Gen-Panel: ID029.01, 8 Gene (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Septooptische Dysplasie *	3 - 5 Wo	E
Gen-Panel: ID378.00, 8 Gene (10,8 kb)		
GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3		
Stickler-Syndrom (STL) *	3 - 5 Wo	E
Gen-Panel: ID062.00, 6 Gene (21,2 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2		
Usher-Syndrom (USH) *	3 - 5 Wo	E
Gen-Panel: ID034.01		
Usher-Syndrom (USH): 13 Gene (70,3 kb)		
ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN		
Usher-Syndrom, Typ I (USH1): 6 Gene (26,2 kb)		
CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G		
Usher-Syndrom, Typ II (USH2): 4 Gene (40,4 kb)		
ADGRV1, PDZD7, USH2A, WHRN		
Usher-Syndrom, Typ III und Typ IV (USH3, USH4): 3 Gene (3,8 kb)		
ARSG, CLRN1, HARS1		

Erkrankung/Diagnostik	Dauer	Material
Vitreoretinopathie *		
Gen-Panel ID352.00		
Vitreoretinopathie: 23 Gene (58,9 kb)		
ATOH7, BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, PAK2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408	3 - 5 Wo	E
Exsudative Vitreoretinopathie (EVR): 15 Gene (34,9 kb)		
ATOH7, CAPN5, CTNNB1, BEST1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RS1, TSPAN12, VCAN, ZNF408		
Syndrome mit Vitreoretinopathie (STL, KNO): 10 Gene (27,6 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, KIF11, NDP, PAK2, RCBTB1		
Walker-Warburg-Syndrom (WWS, MDDGA) *		
Gen-Panel: ID178.00, 14 Gene (23,7 kb)	3 - 5 Wo	E
B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1		
Weill-Marchesani-Syndrom (WMS) *		
Gen-Panel: ID230.00, 4 Gene (20,7 kb)	3 - 5 Wo	E
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD) *		
Gen-Panel: ID101.03, 38 Gene (94,5 kb)	3 - 5 Wo	E
ABCA4, ADAM9, AIPL1, ATF6, CACNA1F, CACNA2D4, CABP4, CDH3, CDHR1, CEP78, CEP250, CFAP418, CNGA3, CNGB3, CNNM4, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RP1, RPGR, RPGRIP1, SEMA4A, TLCD3B, TTLL5, UNC119		
Bindegewebserkrankungen		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik *		
Gen-Panel: ID137.06		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik: 85 Gene (283,3 kb)		
ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ASPH, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, DCC, DLG4, DSE, EFEMP1, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, IPO8, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NKAP, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THBS2, THSD4, TNXB, ZNF469		
Marfan-Syndrom (MFS): 3 Gene (11,8 kb)		
FBN1, TGFBR2, TGFBR1		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1 kb)	4 - 6 Wo	E
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 18 Gene (45,7 kb)		
ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Cutis laxa-Syndrom (ARCL, ADCL): 12 Gene (29,4 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
Stickler-Syndrom (STL): 6 Gene (22,0 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2		
Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Cutis laxa (ARCL, ADCL) *		
Gen-Panel: ID109.03, 13 Gene (27,9 kb)	3 - 5 Wo	E
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1, RIN2		
Ehlers-Danlos-Syndrom (EDS) *		
Gen-Panel: ID039.06		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469	3 - 5 Wo	E
Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 9 Gene (39,6 kb)		
C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1, THBS2		
Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469		
Hereditäre hämorrhagische Teleangiektasie (HHT)		
Gen-Panel: ID155.01, 7 Gene (15,6 kb)	3 - 5 Wo	E
ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4		
Marfan-Syndrom (MFS) *		
Gen-Panel: ID022.00, 3 Gene (11,8 kb)	2 - 4 Wo	E
FBN1 TGFBR1, TGFBR2		
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder *		
Gen-Panel ID194.06		
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 50 Gene (157,9 kb)		
ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP1, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, UPF3B, ZDHHC9, ZNF469		
Marfan-Syndrom (MFS): 3 Gene (11,8 kb)		
FBN1, TGFBR1, TGFBR2	4 - 6 Wo	E
Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 17 Gene (42,1 kb)		
ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Stickler-Syndrom (STL): 6 Gene (21,1 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2		
Syndrome mit marfanoidem Habitus: 20 Gene (52,2 kb)		
CBS, DLG4, EFEMP1, EFEMP2, FBN1, FBN2, NKAP, NPR2, MED12, PLOD1, PRDM5, SKI, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZDHHC9		

Erkrankung/Diagnostik	Dauer	Material
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.08 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 40 Gene (117,6 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (12,6 kb) BGN, IPO8, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit thorakalem Aortenaneurysma: 31 Gene (96,5 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, IPO8, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E
Osteogenesis imperfecta (OI) * Gen-Panel: ID066.02, 21 Gene (39,2 kb) ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1	3 - 5 Wo	E
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (TAA/D) * Gen-Panel: ID020.02 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 17 Gene (44,1 kb) ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	3 - 5 Wo	E
Endokrine Störungen		
Adipositas * Gen-Panel: ID183.03 Adipositas: 57 Gene (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 18 Gene (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 40 Gene (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
Adrenogenitales Syndrom (AGS, CAH) * Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
Diabetes insipidus * Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E
Endokrine Hypertonie * Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E
Glukokortikoid-Mangel (GCCD) * Gen-Panel: ID222.00, 16 Gene (25,1 kb) AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NR0B1, NR3C1, PCSK1, POMC, PROP1, STAR, TBX19, TXNRD2	3 - 5 Wo	E
Hyperaldosteronismus (HALD) * Gen-Panel: ID304.00, 6 Gene (20,6 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5	3 - 5 Wo	E
Hyperinsulinämische Hypoglykämie (HHF) * Gen-Panel: ID126.00, 8 Gene (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hyperparathyreoidismus (HRPT) *	3 - 5 Wo	E
Gen-Panel: ID338.00, 10 Gene (19,2 kb)		
AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung *	3 - 5 Wo	E
Gen-Panel: ID280.00		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung: 44 Gene (85,9 kb)		
ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4		
Glykogenspeicherkrankheit (GSD): 24 Gene (48,4 kb)		
AGL, ALDOA. ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4		
Hyperinsulinämische Hypoglykämie (HHF): 9 Gene (18,8 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A		
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) *	3 - 5 Wo	E
Gen-Panel: ID170.05, 40 Gene (78,9 kb)		
ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11		
Hypoparathyreoidismus *	3 - 5 Wo	E
Gen-Panel: ID353.00, 16 Gene (24,8 kb)		
AIRE, CASR, CYP24A1, FAM111A, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, PTH, SLC34A1, SOX3, STX16, TBCE, TBX1		
Hypophysenadenom (PITA) *	3 - 5 Wo	E
Gen-Panel: ID387.00, 8 Gene (20,7 kb)		
AIP, CDH23, CDKN1B, GNAS, GPR101, MEN1, PRKAR1A, USP8		
Kongenitale Hypothyreose *	3 - 5 Wo	E
Gen-Panel ID369.00		
Kongenitale Hypothyreose: 37 Gene (69,0 kb)		
CDCA8, DIO1, DUOX2, DUOXA2, FOXE1, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, NKX2-1, NKX2-5, OTX2, PAX8, POU1F1, PRKAR1A, PROP1, RNPC3, ROBO1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TUBB1		
Kongenitale Schilddrüsenunterfunktion ohne Kropf (CHNG): 8 Gene (13,0 kb)		
IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR		
Schilddrüsen-Dyshormonogenese (TDH): 8 Gene (23,8 kb)		
DUOX2, DUOXA2, IYD, SLC5A5, SLC26A4, SLC26A7, TG, TPO		
Kombinierter Hypophysenhormonmangel (CPHD): 8 Gene (11,9 kb)		
HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1		
MODY-Diabetes *	3 - 5 Wo	E
Gen-Panel: ID048.01, 14 Gene (22,9 kb)		
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1		
Neonataler Diabetes mellitus *	3 - 5 Wo	E
Panel: ID162.01		
Neonataler Diabetes mellitus: 29 Gene (53,6 kb)		
ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57		
Permanenter neonataler Diabetes mellitus (PNDM): 10 Gene (15,9 kb)		
ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57		
Syndromaler neonataler Diabetes mellitus: 21 Gene (40,5 kb)		
BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5		
Neuroendokrine Neoplasie *	3 - 5 Wo	E
Gen-Panel: ID386.00, 19 Gene (29,2 kb)		
AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL		
Phäochromozytom-Paragangliom-Syndrom (PPGL) *	3 - 5 Wo	E
Gen-Panel: ID042.03, 14 Gene (22,8 kb)		
DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL		
Primäre und prämature Ovarialinsuffizienz (POI, POF) *	3 - 5 Wo	E
Gen-Panel: ID078.06		
Primäre und prämature Ovarialinsuffizienz (POI, POF): 40 Gene (88,5 kb)		
BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, ESR2, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HROB, HSF2BP, INHA, KASH5, LHCGR, MCM8, MCM9, MEIOB, MGA, MRPS22, MSH4, MSH5, NHEJ1, NOBOX, NR5A1, NUP107, POF1B, PSMC3IP, SOHLH1, SPATA22, SPIDR, STAG3, SYCE1, SYCP2L, TP63, XRCC2, ZSWIM7		
Vorzeitige Ovarialinsuffizienz (POF): 26 Gene (65,3 kb)		
BNC1, C14ORF39, DIAPH2, ERCC6, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, KASH5, MCM8, MEIOB, MGA, MSH4, MSH5, NOBOX, NR5A1, POF1B, SPATA22, STAG3, SYCE1, SYCP2L, TP63, XRCC2		
Ovarialdysgenesie (ODG): 11 Gene (19,1 kb)		
BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) *	3 - 5 Wo	E
Gen-Panel: ID250.01		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA): 8 Gene (23,8 kb)		
CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4		
Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb)		
NR3C2, SCNN1A, SCNN1B, SCNN1G		
Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb)		
CUL3, KLHL3, WNK1, WNK4		
Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb)		
SCNN1A, SCNN1B, SCNN1G		
Schilddrüsenkarzinom *	3 - 5 Wo	E
Gen-Panel: ID220.02, 26 Gene (60,5 kb)		
ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53		

Erkrankung/Diagnostik	Dauer	Material
Septooptische Dysplasie * Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
Variante der Geschlechtsentwicklung (DSD) * Gen-Panel ID117.04 Variante der Geschlechtsentwicklung (DSD): 55 Gene (106,2 kb) AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR3C1, NR5A1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX3, SOX8, SOX9, SOX10, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 20 Gene (36,1 kb) ANOS1, AKR1C2, AKR1C4, AR, CBX2, DHH, DHX37, DMRT1, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 7 Gene (8,7 kb) NR2F2, NR5A1, SOX3, SOX9, SRY, WT1, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 41 Gene (80,2 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, NR3C1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX9, SOX10, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
Wachstumshormonmangel (IGHD, CPHD) * Gen-Panel: ID211.02 Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNPC3, OTX2, POU1F1, PROP1, SEMA3A, SOX3 Isolierter Wachstumshormonmangel (IGHD): 5 Gene (6,4 kb) BTK, GH1, GHRHR, GHSR, SOX3 Kombinierter Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A	3 - 5 Wo	E
Entwicklungs- und Wachstumsstörungen		
Adipositas * Gen-Panel: ID183.03 Adipositas: 57 Gene (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 18 Gene (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 40 Gene (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
Autismus-Spektrum-Störungen * Gen-Panel: ID076.04, 168 Gene (664,6 kb) AHDC1, ADNP, ADSL, AFF2, AGO1, ALDH5A1, ANK2, ANKRD11, ARHGEF9, ARID1B, ARX, ASH1L, ASTN2, ASXL3, ATP1A1, AUTS2, BAZ2B, BCKDK, BCL11A, BRSK2, CACNA1C, CAPRIN1, CASK, CASZ1, CC2D1A, CDKL5, CELF4, CHD2, CHD7, CHD8, CIC, CNOT3, CNTN4, CNTNAP2, CREBBP, CSDE1, CSNK2A1, CTNNB1, CUL3, DDX3X, DEAF1, DHCR7, DIP2A, DLGAP2, DNMT3A, DPP6, DSCAM, DYNC1H1, DYRK1A, EBF3, EHMT1, EIF4E, EP300, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, GIGYF1, GLRA2, GRIA2, GRIN2A, GRIN2B, HERC2, HNRNPU, IL1RAPL1, IQSEC2, IRF2BPL, KATNAL2, KCNQ3, KDM5A, KDM5C, KDM6B, KMT2A, KMT2C, KMT2E, KMT5B, MAGEL2, MAOA, MBD5, MBOAT7, MECP2, MED13, MED13L, MEF2C, MEIS2, MYT1L, NAA15, NEXMIF, NF1, NHS, NIPBL, NLGN1, NLGN3, NLGN4X, NOVA2, NR1I3, NR4A2, NRXN1, NRXN2, NRXN3, NSD1, OPHN1, PAH, PAX5, PCDH19, PDZD8, PHF12, PHF2, PHF21A, PHF3, POGZ, PPP2R5D, PSMD12, PTCHD1, PTEN, RAB39B, RAI1, RELN, RERE, RFX3, RIMS1, RORB, RPL10, SATB2, SCN1A, SCN2A, SEMA5A, SETD2, SETD5, SGSH, SHANK2, SHANK3, SLC6A1, SLC6A8, SLC9A6, SLC9A9, SMARCB1, SMARCC2, SON, SOX5, SPAST, STXBP1, SYN1, SYNGAP1, TANC2, TBL1XR1, TBR1, TCF20, TCF4, TLK2, TMLHE, TRIP12, TRPC6, TRRAP, TSC1, TSC2, UBE3A, UPF3B, VAMP2, VPS13B, WAC, WDFY3, ZMYM3, ZMYND8, ZNF292, ZNF462, ZSWIM6	4 - 6 Wo	E
Coffin-Siris-Syndrom (CSS) * Gen-Panel: ID118.01, 14 Gene (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 Wo	E
Cornelia-de-Lange-Syndrom (CDLS) * Gen-Panel: ID033.02, 8 Gene (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 Wo	E
FG-Syndrom (FGS) * Gen-Panel: ID215.00, 3 Gene (17,2 kb) CASK, FLNA, MED12	3 - 5 Wo	E
Großwuchs, umfassende Diagnostik * Gen-Panel: ID299.01, 64 Gene (211,5 kb) ABCC9, AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, FBN1, FIBP, GFAP, GLI3, GPC3, GPC4, GRIA3, H1-4, HEPACAM, HERC1, HUWE1, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIGA, PIK3CA, PIK3R2, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SHANK, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMEM94, TRIP12, UPF3B, ZBTB20, ZDHHC9	4 - 6 Wo	E
Großwuchssyndrome (SOTOS, BWS) * Gen-Panel: ID073.05 Großwuchssyndrome (SOTOS, BWS): 15 Gene (41,2 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, PDGFRB, PTEN, SETD2, SUZ12, RNF125 Sotos-ähnliche Großwuchssyndrome: 6 Gene (23,1 kb) EED, EZH2, NFIX, NSD1, SETD2, SUZ12 Beckwith-Wiedemann-ähnliche Großwuchssyndrome: 5 Gene (10,1 kb) CDKN1C, DIS3L2, GPC3, GPC4, OFD1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *	3 - 5 Wo	E
Gen-Panel: ID292.00, 6 Gene (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD, IDD) *	4 - 6 Wo	E
Gen-Panel: ID036.06		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD, IDD): 105 Gene (354,6 kb)		
ACTL6B, ADNP, AHDC1, AP2M1, ARID1A, ARID1B*, ARID2, ASH1L, ATP2B1, AUTS2, BCL11A, BCL11B, BICRA, BRPF1, CACNG2, CAMK2A, CAMK2B, CAMK2G, CCNK, CDH15, CDK8, CERT1, CHAMP1, CHD3, CHD8, CIC, CLTC, CNOT2, CNOT3, CTCF, CTNNB1, DDX6, DEAF1, DHX9, DLG4, DPF2, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, FBXO11, FOXP1, GATAD2B, GNB1, GRIA1, GRIN1, GRIN2B, HIVEP2, HNRNPC, KAT6A, KCNQ5, KDM3B, KDM4B, KIF1A, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MEF2C, MTSS2, MYT1L, NAA15, NR4A2, NUS1, PACS1, PAK1, PHF21A, POGZ, PPP2R1A, PPP2R5D, PURA*, RAC1, RFX7, RORA, SET, SETBP1, SETD1B, SETD2, SETD5, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4, SOX6, SRRM2, STAG1, SYNGAP1, TAF4, TANC2, TBL1XR1, TBR1, TCP1, TLK2, TNPO2, TRIO, TRIP12, USP7, ZBTB18, ZMYND11, ZNF292		
Intellektuelle Entwicklungsstörung, autosomal-dominant, nicht-syndromal (MRD): 62 Gene (224,4 kb)		
AP2M1, ARID1A, ARID1B, ASH1L, ATP2B1, AUTS2, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CIC, CLTC, CTCF, DEAF1, DHX9, DLG4, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN2B, HNRNPC, HIVEP2, KCNQ5, KDM4B, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, RAC1, RFX7, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SOX11, SRRM2, STAG1, SYNGAP1, TAF4, TBL1XR1, TLK2, TRIO, ZBTB18, ZMYND11, ZNF292		
Intellektuelle Entwicklungsstörung, autosomal-dominant, syndromal (IDD): 55 Gene (168,0 kb)		
ACTL6B, ADNP, AHDC1, ARID1A, ARID1B, ARID2, BCL11A, BCL11B, BICRA, BRPF1, CCNK, CDK8, CERT1, CHAMP1, CHD3, CHD8, CNOT2, CNOT3, CTNNB1, DDX6, DEAF1, DPF2, FBXO11, FOXP1, GATAD2B, GRIN1, KAT6A, KDM3B, KIF1A, MEF2C, MTSS2, NR4A2, PACS1, PAK1, PHF21A, POGZ, PPP2R1A, PPP2R5D, PURA, RORA, SETD1B, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4, SOX6, TANC2, TBR1, TCP1, TNPO2, TRIP12, USP7		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT, IDD) *	4 - 6 Wo	E
Gen-Panel: ID037.03		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT, IDD): 77 Gene (201,7 kb)		
ABCA2, ACTL6B, ADAT3, ALG14, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CPE, CRADD, CRBN, DEAF1, EDC3, EIF3F, ELP2, FBXL3, FBXO31, FERRY3, FMN2, GNB5, GRIA1, GRIK2, HERC2, HNMT, IMPA1, IQSEC1, KDM5B, KPTN, LGI3, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NEMF, NSUN2, NSUN6, NUDT2, OTUD6B, PDE2A, PDZD8, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, PUS7, RSRC1, RUSC2, SCAPER, SLC45A1, SLC6A17, ST3GAL3, TAF13, TAF2, TECR, TMEM94, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, WIPI2, ZBTB11, ZC3H14		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv, nicht-syndromal (MRT): 58 Gene (157,9 kb)		
ADAT3, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31, FERRY3, FMN2, GRIA1, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NSUN2, NSUN6, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, ZBTB11, ZC3H14		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv, syndromal (IDD): 20 Gene (48,5 kb)		
ABCA2, ALG14, CPE, DEAF1, FBXL3, GNB5, IQSEC1, LGI3, NEMF, NUDT2, OTUD6B, PDE2A, PDZD8, PUS7, SCAPER, SLC45A1, TAF2, TMEM94, WIPI2, ZBTB11		
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS) *	4- 6 Wo	E
Gen-Panel: ID038.06		
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS): 77 Gene (194,6 kb)		
ACSL4, AFF2, AP1S2, ARX, ATP6AP2, ATRX, BRWD3, CASK, CLCN4, CLIC2, CNKSR2, CSTF2, CUL4B, DDX3X, DLG3, EIF2S3, FAM50A, FGD1, FGF13, FMR1, FRMPD4, FTSJ1, GDI1, GLRA2, GRIA3, HCFC1, HNRNPH2, HS6ST2, HUWE1, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIF4A, KLHL15, LAS1L, MECP2, MED12, MID2, MSL3, NEXMIF, NKAP, NONO, OGT, OPHN1, PAK3, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, RPS6KA3, SLC16A2, SLC9A6, SLC9A7, SLITRK2, SMS, STEEP1, SYN1, SYP, TAF1, THOC2, TSPAN7, UBE2A, UPF3B, USP9X, USP27X, WNK3, ZMYM3, ZC4H2, ZDHHC9, ZFX, ZNF711		
Intellektuelle Entwicklungsstörung, X-chromosomal, nicht-syndromal (XLID, MRX): 29 Gene (77,8 kb)		
ACSL4, AFF2, ARX, BRWD3, DLG3, FGF13, FRMPD4, FTSJ1, GDI1, HCFC1, IL1RAPL1, IQSEC2, KIF4A, KLHL15, MID2, NEXMIF, OGT, PAK3, RAB39B, RPS6KA3, SLC9A7, STEEP1, SYN1, SYP, THOC2, TSPAN7, USP9X, USP27X, ZNF711		
Intellektuelle Entwicklungsstörung, X-chromosomal, syndromal (MRXS): 52 Gene (130,8 kb)		
AFF2, AP1S2, ARX, ATP6AP2, ATRX, CASK, CLCN4, CLIC2, CNKSR2, CSTF2, CUL4B, DDX3X, EIF2S3, FAM50A, FGD1, FMR1, GLRA2, GRIA3, HNRNPH2, HS6ST2, HUWE1, IGBP1, KDM5C, LAS1L, MECP2, MED12, MSL3, NKAP, NONO, OPHN1, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, SLC16A2, SLC9A6, SLITRK2, SMS, TAF1, UBE2A, UPF3B, USP9X, ZC4H2, WNK3, ZMYM3, ZDHHC9, ZFX		
Intellektuelle Entwicklungsstörung und Makrozephalie *	4 - 6 Wo	E
Gen-Panel: ID131.03		
Intellektuelle Entwicklungsstörung und Makrozephalie: 48 Gene (147,3 kb)		
ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Makrozephalie: 21 Gene (70,8 kb)		
CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Makrozephalie: 10 Gene (20,2 kb)		
ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMCO1, ZBTB7A		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Makrozephalie: 18 Gene (57,9 kb)		
BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9		
Intellektuelle Entwicklungsstörung und Mikrozephalie *	4 - 6 Wo	E
Gen-Panel: ID129.02		
Intellektuelle Entwicklungsstörung und Mikrozephalie: 80 Gene (229,8 kb)		
ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNNB1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNPH2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Mikrozephalie: 32 Gene (100,5 kb)		
AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Mikrozephalie: 21 Gene (41,0 kb)		
ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Mikrozephalie: 27 Gene (88,3 kb)		
ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2		

Erkrankung/Diagnostik	Dauer	Material
Kabuki-Syndrom (KABUK) * Gen-Panel: ID127.00, 2 Gene (20,8 kb) KDM6A, KMT2D	3 - 5 Wo	E
Kleinwuchs, umfassende Diagnostik * Gen-Panel ID340.02 Kleinwuchs, umfassende Diagnostik: 207 Gene (511,0 kb) ACAN, ACP5, ACTB, ACTG1, AGPS, ALPL, AMMECR1, ANKRD11, ARCN1, ARSB, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BLM, BMP2, BMPR1B, BRAF, BRCA1, BRCA2, BRIP1, BTK, CBL, CCDC8, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CFAP410, COL10A1, COL11A2, COL27A1, COL2A, COMP, CREBBP, CRIPT, CSGALNACT1, CUL7, DDR2, DDRGK1, DHCR7, DNA2, DONSON, DPH1, EP300, ERCC4, ERCC6, ERCC8, EXOC6B, EXOSC2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGD1, FGFR1, FGFR3, FLNB, FN1, GALNS, GDF5, GH1, GHR, GHRHR, GHSR, GLB1, GLI2, GMNN, GNPAT, GPX4, GRHL2, GSC, GUSB, GZF1, HDAC8, HESX1, HMGA2, HRAS, HYAL1, IDUA, IGF1, IGF1R, IGF2, IGFALS, INSR, IRS1, IRS4, KIF22, KMT2A, KRAS, LARP7, LFNG, LHX3, LHX4, LTBP3, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MCM5, MRAS, NBAS, NBN, NEPRO, , NIPBL, NKX2-5, NKX3-2, NOTCH2, NPR2, NRAS, NSMCE2, OBSL1, ORC1, ORC4, ORC6, OTX2, PALB2, PAM16, PAPPA2, PAX8, PCNT, PEX5, PEX7, PHEX, PIK3R1, PISD, PLAG1, PLCB3, POC1A, POLR3GL, POP1, POU1F1, PPP1CB, PPP1R15B, PPP3CA, PRKG2, PRMT7, PROP1, PTH1R, PTPN11, PUS7, RAD21, RAD51, RAD51C, RAF1, RBBP8, RFWD3, RIT1, RMRP, RNPC3, RNU4ATAC, ROBO1, RPL13, RRAS2, RSPRY1, RTTN, SCUBE3, SGMS2, SHOC2, SHOX, SLC10A7, SLC26A2, SLX4, SMARCA2, SMC1A, SMC3, SOS1, SOS2, SOX, SOX3, SPRED2, SRCAP, STAT5B, TALDO1, TBCE, TBL1X, TBX15, TBX19, TBX2, THRA, TKT, TONSL, TOP3A, TRAIP, TRAPPC2, TRHR, TRIM37, TRIP11, TRMT10A , TRPV4, TSHB, TSHR, UBE2T, XRCC2, XRCC4 Wachstumshormonmangel (IGHD, CPHD): 14 Gene (23,0 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1, SOX3 Noonan-Syndrom (NS): 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Meier-Gorlin-Syndrom (MGORS): 8 Gene (12,6 kb) CDC45, CDC6, CDT1, GMNN, MCM5, ORC1, ORC4, ORC Seckel-Syndrom (SCKL): 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP Kongenitale Hypothyreose (CHNG): 8 Gene (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Skelettdysplasie (SED, SMD, AMD): 35 Gene (83,9 kb) ACAN, B3GALT6, BGN, BMPR1B, CFAP410, COL11A2, COL2A1, COMP, DDRGK1, DDR2, EXOC6B, FGFR3, FN1, GDF5, GPX4, KIF22, NEPRO, NKX3-2, NPR2, PAM16, PAPSS2, PISD, PLCB3, RMRP, RNU4ATAC, POP1, PRKG2, RPL13, RSPRY1, SIK3, SLC26A2, TONSL, TRAPPC2, TRIP11, TRPV4	4 - 6 Wo	E
Komplexe neurologische Entwicklungsstörungen (NED) * Gen-Panel: ID358.01, 193 Gene (496,0 kb) ACBD6, ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BAZ2B, BCAS3, BPTF, BRAT1, C18ORF32, CAPRIN1, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CERT1, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DPH5, DYNC1I2, EEF1D, EMC10, ESAM, EXOC2, EXOC7, EXOC8, FAM177A1, FBXW11, FDFT1, FEM1B, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, GTPBP2, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HDAC4, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCND1, KCNN2, KDM5A, KDM6B, LNPK, MADD, MAP4K4, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NAV3, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPMT1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, RNU4-2, SARS1, SEC31A, SEL1L, SETD1A, SHMT2, SHQ1, SLC4A10, SMG8, SMG9, SMPD4, SNF8, SNIP1, SPOP, SPOUT1, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, TTI1, UBE3C, UBE4A, UBR5, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZBTB11, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 Wo	E
Noonan-Syndrom (NS) * Gen-Panel: ID023.06, 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 Wo	E
Pitt-Hopkins-Syndrom (PTHS) * Gen-Panel: ID106.00, 3 Gene (10,6 kb) CNTNAP2, NRXN1, TCF4	3 - 5 Wo	E
Progerie und progeroide Syndrome * Gen-Panel: ID147.01, 25 Gene (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 Wo	E
RASopathien * Gen-Panel: ID015.05 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan-Syndrom (NS): 15 Gene (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 Wo	E
Rett-Syndrom (RTT) und ähnliche Krankheitsbilder * Gen-Panel: ID125.01, 16 Gene (45,3 kb) CDKL5, FOXG1, GABBR2, GABRA2, GABRB2, GRIN2B, HTT, IQSEC2, MECP2, MEF2C, NTNG2, SHANK3, STXBP1, TCF4, UBE3A, WDR45	3 - 5 Wo	E
Rubinstein-Taybi-Syndrom (RSTS) * Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
Sprachentwicklungsstörung (DLD, CAS) * Gen-Panel: ID368.00 Sprachentwicklungsstörung (DLD, CAS): 34 Gene (119,7 kb) ATP2C2, BCL11A, BUD13, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, NFXL1, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TM4SF20, TNRC6B, UPF2, WDR5, ZFHX4, ZNF142, ZNF277 Kindliche Sprechapraxie (CAS): 29 Gene (110,2 kb) BCL11A, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TNRC6B, UPF2, WDR5, ZFHX4, ZNF142 Spezifische Sprachbeeinträchtigung (SLI): 5 Gene (9,5 kb) TP2C2, BUD13, NFXL1, TM4SF20, ZNF277	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Seckel-Syndrom (SCKL) *		
Gen-Panel: ID113.00, 9 Gene (33,5 kb)	3 - 5 Wo	E
ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP		
Wachstumshormonmangel (IGHD, CPHD) *		
Gen-Panel: ID211.02		
Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb)		
BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNPC3, OTX2, POU1F1, PROP1, SEMA3A, SOX3	3 - 5 Wo	E
Isolierter Wachstumshormonmangel (IGHD): 5 Gene (6,4 kb)		
BTK, GH1, GHRHR, GHSR, SOX3		
Kombinierter Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb)		
GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A		
Wachstumsstörung und Makrozephalie *		
Gen-Panel: ID072.03		
Wachstumsstörung und Makrozephalie: 30 Gene (80,9 kb)		
AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12	3 - 5 Wo	E
Großwuchssyndrom und Makrozephalie: 11 Gene (33,0 kb)		
DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12		
Noonan-Syndrom und Makrozephalie: 10 Gene (15,2 kb)		
BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1		
3M-Syndrom *		
Gen-Panel: ID214.00, 3 Gene (12,4 kb)	3 - 5 Wo	E
CCDC8, CUL7, OBSL1		
Epilepsien und Migräne		
Absence-Epilepsie (EJA, ECA) *		
Gen-Panel: ID057.02, 10 Gene (21,8 kb)	3 - 5 Wo	E
CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, KCNMA1, RORB, SLC2A1, SLC12A5		
Benigne neonatale und infantile Krampfanfälle (BFNS, BFIS) *		
Gen-Panel: ID134.01, 6 Gene (19,8 kb)	3 - 5 Wo	E
CHRNA2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A		
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE) *		
Gen-Panel ID080.03		
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb)		
AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG	4 - 6 Wo	E
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), autosomal-dominant: 51 Gene (143,1 kb)		
ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CDK19, CELF2, CHD2, CUX2, CYFIP2, DNM1, EEF1A2, FBXO28, FGF12, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GNAO1, GRIN2B, GRIN2D, HCN1, HNRNPU, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, NEUROD2, NSF, NTRK2, PACS2, PHACTR1, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN2A, SCN3A, SCN8A, SIK1, SLC1A2, SPTAN1, STXBP1, YWHAG		
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), autosomal-rezessiv: 45 Gene (105,0 kb)		
AARS1, ACTL6B, ADAM22, AP3B2, ARV1, CAD, CNPY3, CPLX1, DALRD3, DENND5A, DMXL2, DOCK7, FRRS1L, GAD1, GLS, GOT2, GRIN1, GUF1, HID1, ITPA, MDH1, MDH2, NECAP1, PARS2, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, SCN1B, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC38A3, ST3GAL3, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX		
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), X-chromosomal: 9 Gene (20,3 kb)		
ALG13, ARHGEF9, ARX, CDKL5, FGF13, PCDH19 PIGA, SLC35A2, SMC1A		
Epilepsien, umfassende Diagnostik *		
Gen-Panel ID061.05		
Epilepsien, umfassende Diagnostik: 163 Gene (388,9 kb)		
AARS1, ACTL6B, ADAM22, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ASAH1, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, BRAT1, CACNA1A, CACNA1E, CACNB4, CAD, CASR, CDK19, CDKL5, CELF2, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CILK1, CLCN2, CLN8, CNPY3, CNTN2, CNTNAP2, CPA6, CPLX1, CSTB, CUX2, CYFIP2, DALRD3, DENND5A, DEPDC5, DMXL2, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GLS, GNAO1, GOT2, GOSR2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GUF1, HCN1, HCN2, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC1, KCNC2, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LGI1, LMNB2, MDH1, MDH2, MECP2, MEF2C, MTOR, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PLPBP, PPP3CA, PRDM8, PRICKLE1, PRRT2, RELN, RHOBTB2, RNF13, ROGDI, RORA, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC38A3, SLC6A1, SLC7A60S, SMC1A, SNIP1, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG		
Fokale Epilepsien: 17 Gene (50,9 kb)	4 - 6 Wo	E
CHRNA2, CHRNA4, CHRNB2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24		
Generalisierte Epilepsien: 39 Gene (72,6 kb)		
ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A60S, SLC12A5, STX1B, TBC1D24		
Epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb)		
AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP*, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG		

Erkrankung/Diagnostik	Dauer	Material
Epilepsie mit schwerer Entwicklungsstörung *	3 - 5 Wo	E
Gen-Panel: ID060.00, 20 Gene (68,4 kb)		
ARHGEF9, ARX, CACNA1A, CDKL5, FOXG1, KCNQ2, MECP2, MEF2C, MTHFR, PCDH19, SCN1A, SCN2A, SCN8A, SCN9A, SLC25A22, SLC2A1, SPTAN1, STXBP1, SYNGAP1, UBE3A		
Epileptische Enzephalopathien, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel ID047.04		
Epileptische Enzephalopathien, umfassende Diagnostik: 163 Gene (371,9 kb)		
AARS1, ABAT, ACTL6B, ADAM22, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP3B2, ARHGEF9, ARV1, ARX, ASNS, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, BRAT1, BSCL2, BTD, CACNA1A, CACNA1E, CAD, CCDC88A, CDK19, CDKL5, CELF2, CHD2, CLCN4, CNPY3, CPLX1, CNTNAP2, CUX2, CYFIP2, D2HGDH, DALRD3, DENND5A, DNM1, DNM1L, DMXL2, DOCK7, EEF1A2, ETHE1, FGF12, FGF13, FOLR1, FOXG1, FBXO28, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GAMT, GCSH, GLDC, GLS, GNAO1, GOT2, GPHN, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, IDH2, IFIH1, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, LIAS, MECP2, MEF2C, MDH1, MDH2, MFF, MOCS1, MOCS2, MTHFR, NAXD, NAXE, NECAP1, NEUROD2, NRXN1, NSF, NTRK2, PACS2, PARS2, PC, PCDH19, PHACTR1, PHGDH, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PPP3CA, PURA, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SERPINI1, SIK1, SLC1A2, SLC2A1, SLC12A5, SLC13A5, SLC6A8, SLC6A9, SLC9A6, SLC19A3, SLC25A1, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TCF4, TPK1, TRAK1, TREX1, UBA5, UGDH, UGP2, WDR45, WWOX, YWHAG		
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb)		
AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG		
Metabolische Enzephalopathie mit Epilepsie: 29 Gene (44,8 kb)		
ABAT, ADSL, ALDH5A1, ALDH7A1, AMT, BTD, D2HGDH, FOLR1, GAMT, GCSH, GLDC, GPHN, IDH2, LIAS, MDH2, MOCS1, MOCS2, MTHFR, PC, PHGDH, PNPO, POLG, SLC1A2, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, TPK1		
Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFSP) *	3 - 5 Wo	E
Gen-Panel: ID059.03		
Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFSP): 9 Gene (35,9 kb)		
ADGRV1, CPA6, GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B		
Fieberkrämpfe (FEB): 5 Gene (30,3 kb)		
ADGRV1, CPA6, GABRG2, HCN2, SCN1A		
Generalisierte Epilepsie mit Fieberkrämpfen (GEFSP): 7 Gene (15,7 kb)		
GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B		
Fokale Epilepsien *	3 - 5 Wo	E
Gen-Panel: ID208.01		
Fokale Epilepsien: 17 Gene (50,9 kb)		
CHRNA2, CHRNA4, CHRNB2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24		
Fokale Epilepsie mit variablen Herden (FFEVF): 4 Gene (13,7 kb)		
DEPDC5, NPRL2, NPRL3, SCN3A		
Nächtliche Frontallappenepilepsie (ENFL): 5 Gene (13,5 kb)		
CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNT1		
Temporallappenepilepsie (ETL): 4 Gene (13,4 kb)		
CPA6, GAL, LGI1, RELN		
Generalisierte Epilepsien *	3 - 5 Wo	E
Gen-Panel: ID040.05		
Generalisierte Epilepsien: 39 Gene (72,6 kb)		
ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A60S, SLC12A5, STX1B, TBC1D24		
Myoklonusepilepsie (EJM, EPM): 24 Gene (45,3 kb)		
ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6B, SLC7A60S, TBC1D24		
Absence-Epilepsie (EJA, ECA): 9 Gene (18,3 kb)		
CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, RORB, SLC2A1, SLC12A5		
Generalisierte Epilepsie mit Fieberkrämpfen (GEFSP): 7 Gene (15,7 kb)		
GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B		
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFSP, GEFS+) *	3 - 5 Wo	E
Gen-Panel: ID235.03, 8 Gene (17,2 kb)		
GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, SLC32A1, STX1B		
Hemiplegische Migräne (FHM) *	3 - 5 Wo	E
Gen-Panel: ID064.02, 7 Gene (23,1 kb)		
ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1		
Migräne (MGR) *	3 - 5 Wo	E
Gen-Panel: ID065.01		
Migräne (MGR): 16 Gene (44,8 kb)		
ALPK1, ATP1A2, ATP1A3, CACNA1A, CSNK1D, ESR1, HTRA1, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1, TNF, TREX1		
Familiäre hemiplegische Migräne (FHM): 3 Gene (16,6 kb)		
ATP1A2, CACNA1A, SCN1A		
Myoklonusepilepsie (EPM, EJM) *	3 - 5 Wo	E
Gen-Panel: ID116.03		
Myoklonusepilepsie (EJM, EPM): 24 Gene (45,3 kb)		
ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6B, SLC7A60S, TBC1D24		
Juvenile Myoklonusepilepsie (EJM): 12 Gene (28,0 kb)		
CACNB4, CASR, CILK1, CLCN2, EFHC1, GABRA1, GABRD, RORB, SCN1A, SLC2A1, SLC12A5, TBC1D24		
Progressive Myoklonusepilepsie (EPM): 16 Gene (24,0 kb)		
ASAH1, CERS1, CLN8, CSTB, EPM2A, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SEMA6B, SLC7A60S		

Erkrankung/Diagnostik	Dauer	Material
Metabolische Epilepsien * Gen-Panel: ID303.01 Metabolische Epilepsien: 84 Gene (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycin-Enzephalopathie (GCE): 5 Gene (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdän-Cofaktor-Defizienz (MOCOD): 3 Gene (3,7 kb) GPHN, MOCS1, MOCS2 Zerebrales Kreatinmangelsyndrom (CCDS): 3 Gene (3,9 kb) GAMT, GATM, SLC6A8 Ahornsirupkrankheit (MSUD): 5 Gene (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxy-Glutarazidurie: 3 Gene (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-Gangliosidose: 3 Gene (3,8 kb) HEXA, HEXB, GM2A Neuronale Ceroid-Lipofuszinose (CLN): 12 Gene (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 Wo	E
Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter * Gen-Panel: ID171.00, 18 Gene (31,5 kb) ALDH5A1, ATP7A, BTD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Schul- und Jugendalter * Gen-Panel: ID172.00, 15 Gene (28,1 kb) ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Neugeborenenalter * Gen-Panel: ID135.00, 25 Gene (39,4 kb) ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO	3 - 5 Wo	E
Fertilitätsstörungen		
Adrenogenitales Syndrom (AGS, CAH) * Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
Azoospermie und Kryptozoospermie * Gen-Panel: ID391.01, 60 Gene (142,0 kb) ADAD2, ADGRG2, AR, C14ORF39, CATIP, CATSPER1, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, FANCM, FKBP6, GCNA, HFM1, KASH5, KCTD19, KLHL10, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PMFBP1, PNLDC1, RAD21L1, RBBP7, RNF212, RPL10L, SHOC1, SOHLH1, SPATA22, SPINK2, SPO11, STAG3, STRA8, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TERB1, TERB2, TEX11, TEX14, TEX15, USP9Y, XRCC2, ZMYND15, ZSWIM7	4 - 6 Wo	E
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) * Gen-Panel: ID170.05, 40 Gene (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 Wo	E
Männliche Infertilität, umfassende Diagnostik * Gen-Panel ID192.07 Männliche Infertilität, umfassende Diagnostik: 135 Gene (446,9 kb) ACR, ACTL7A, ACTL9, ADAD2, ADGRG2, AK7, AK9, AKAP3, AR, ARMC12, ARMC2, ATG4D, AURKC, BRDT, C14ORF39, CATIP, CATSPER1, CATSPER2, CATSPERT, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP206, CFAP251, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH12, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, GPAT2, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRRC23, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PLD6, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RAD21L1, RBBP7, RNF212, RPL10L, SEPTIN4, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG6, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SPO11, SSX1, STAG3, STK33, STRA8, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TDRD12, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Störung der Spermatogenese (SPGF): 112 Gene (395,3 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, ATG4D, AURKC, BRDT, C14ORF39, CATIP, CATSPER1, CATSPERT, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP206, CFAP251, CT55, CYLC1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH12, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, IFT74, IQCN, KASH5, KCNU1, KLHL10, LRRC23, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RBBP7, RNF212, RPL10L, SEPTIN4, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Obstruktive Azoospermie (CBAVD, OAZON): 3 Gene (8,2 kb) ADGRG2, CFTR, CLDN2	4 - 6 Wo	E
Multiple morphologische Anomalien der Spermienflagellen (MMAF) * Gen-Panel: ID390.00, 37 Gene (213,9 kb) AK7, AKAP3, ARMC2, CCDC34, CCDC146, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DRC1, DZIP1, FSIP2, IFT74, QRICH2, SPEF2, SSX1, STK33, TTC21A, TTC29, USP26, WDR19	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Ovarialdysgenesie (ODG) *	3 - 5 Wo	E
Gen-Panel: ID293.02		
Ovarialdysgenesie (ODG): 18 Gene (30,9 kb)		
BMP15, CLPP, DAP3, ERAL1, ESR2, FSHR, HARS2, HROB, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7		
XX-Gonadendysgenesie (ODG): 11 Gene (19,1 kb)		
BMP15, ESR2, FSHR,HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7		
XX-Gonadendysgenesie mit Taubheit (PRLTS): 7 Gene (11,8 kb)		
CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, TWNK		
Perrault-Syndrom (PRLTS) *	3 - 5 Wo	E
Gen-Panel: ID388.00, 9 Gene (14,1 kb)		
CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, MRPL49, PRORP, TWNK		
Primäre und prämature Ovarialinsuffizienz (POI, POF) *	3 - 5 Wo	E
Gen-Panel: ID078.06		
Primäre und prämature Ovarialinsuffizienz (POI, POF): 40 Gene (88,5 kb)		
BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, ESR2, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HROB, HSF2BP, INHA, KASH5, LHCGR, MCM8, MCM9, MEIOB, MGA, MRPS22, MSH4, MSH5, NHEJ1, NOBOX, NR5A1, NUP107, POF1B, PSMC3IP, SOHLH1, SPATA22, SPIDR, STAG3, SYCE1, SYCP2L, TP63, XRCC2, ZSWIM7		
Vorzeitige Ovarialinsuffizienz (POF): 26 Gene (65,3 kb)		
BNC1, C14ORF39, DIAPH2, ERCC6, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, KASH5, MCM8, MEIOB, MGA, MSH4, MSH5, NOBOX, NR5A1, POF1B, SPATA22, STAG3, SYCE1, SYCP2L, TP63, XRCC2		
Ovarialdysgenesie (ODG): 11 Gene (19,1 kb)		
BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7		
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *	4 - 6 Wo	E
Gen-Panel: ID085.03, 50 Gene (164,8 kb)		
BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10		
Störung der Oozyten-, Zygoten- und Embryonen-Reifung (OZEMA) *	3 - 5 Wo	E
Gen-Panel: ID239.03, 26 Gene (46,2 kb)		
ASTL, BTG4, CDC20, CHEK1, FBXO43, KHDC3L, KPNA7, MEI1, MOS, NLRP2, NLRP5, NLRP7, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TOP6BL, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3		
Variante der Geschlechtsentwicklung (DSD) *	4 - 6 Wo	E
Gen-Panel ID117.04		
Variante der Geschlechtsentwicklung (DSD): 55 Gene (106,2 kb)		
AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR3C1, NR5A1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX3, SOX8, SOX9, SOX10, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2		
46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 20 Gene (36,1 kb)		
ANOS1, AKR1C2, AKR1C4, AR, CBX2, DHH, DHX37, DMRT1, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2		
46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 7 Gene (8,7 kb)		
NR2F2, NR5A1, SOX3, SOX9, SRY, WT1, WNT4		
Störung der Geschlechtsentwicklung (DSD), syndromal: 41 Gene (80,2 kb)		
AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, NR3C1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX9, SOX10, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1		
Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb)		
CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR		
Weibliche Infertilität, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel ID389.00		
Weibliche Infertilität, umfassende Diagnostik: 80 Gene (156,6 kb)		
ASTL, BMP15, BNC1, BTG4, C14ORF39, CDC20, CHEK1, CLPP, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DAP3, DIAPH2, ERAL1, ERCC6, ESR2, FANCM, FBXO43, FIGLA, FMR1, FOXL2, FSHR, GDF9, HARS2, HFM1, HROB, HSD17B4, HSD3B2, HSF2BP, INHA, KASH5, KHDC3L, KPNA7, LARS2, LHCGR, MCM8, MCM9, MEI1, MEIOB, MGA, MOS, MRPS22, MSH4, MSH5, NHEJ1, NLRP2, NLRP5, NLRP7, NOBOX, NR5A1, NUP107, PABPC1L, PADI6, PANX1, PATL2, POF1B, POR, PSMC3IP, REC114, SOHLH1, SPATA22, SPIDR, STAG3, STAR, SYCE1, SYCP2L, TLE6, TOP6BL, TP63, TRIP13, TUBB8, TWNK, WEE2, XRCC2, ZFP36L2, ZP1, ZP2, ZP3, ZSWIM7		
Primäre und prämature Ovarialinsuffizienz (POI, POF): 40 Gene (88,5 kb)		
BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, ESR2, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HROB, HSF2BP, INHA, KASH5, LHCGR, MCM8, MCM9, MEIOB, MGA, MRPS22, MSH4, MSH5, NHEJ1, NOBOX, NR5A1, NUP107, POF1B, PSMC3IP, SOHLH1, SPATA22, SPIDR, STAG3, SYCE1, SYCP2L, TP63, XRCC2, ZSWIM7		
Störung der Oozyten-, Zygoten- und Embryonenreifung (OZEMA): 26 Gene (46,2 kb)		
ASTL, BTG4, CDC20, CHEK1, FBXO43, KHDC3L, KPNA7, MEI1, MOS, NLRP2, NLRP5, NLRP7, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TOP6BL, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3		
Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb)		
CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR		
Zystische Fibrose (CF) *	3 - 5 Wo	E
Gen-Panel: ID045.00, 1 Gen (4,4 kb)		
CFTR		

Erkrankung/Diagnostik	Dauer	Material
Fettstoffwechselstörungen		
Fettstoffwechselstörungen, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID044.03, 62 Gene (114,6 kb)		
ABCA1, ABCG5, ABCG8, ACADM, ACADS, ACADVL, ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA1, APOA2, APOA4, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CETP, CIDEC, CREB3L3, CYP27A1, CYP7A1, DHCR7, DHCR24, GCKR, GK, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIMA1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPA, LPL, MTTP, NPC1, NPC1L1, NPC2, PCSK9, PCYT1A, PLAAT3, PLIN1, PNPLA5, POLD1, PPARG, SAR1B, SCARB1, SMPD1, SORT1, ZMPSTE24		
Fettstoffwechselstörung durch LDL-Mangel *	3 - 5 Wo	E
Gen-Panel: ID094.01, 10 Gene (29,6 kb)		
ANGPTL3, ANGPTL4, ANGPTL8, APOB, APOE, LIMA1, MTTP, NPC1L1, PCSK9, SAR1B		
HDL-assoziierte Fettstoffwechselstörung *	3 - 5 Wo	E
Gen-Panel: ID096.01, 15 Gene (25,5 kb)		
ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA2, APOA4, APOC3, CETP, LCAT, LIPC, LIPG, NPC1, NPC2, SCARB1, SMPD1		
Hypercholesterinämie *	3 - 5 Wo	E
Gen-Panel: ID024.02, 13 Gene (36,4 kb)		
ABCG5, ABCG8, APOB, APOE, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1		
Hypertriglyzeridämie *	3 - 5 Wo	E
Gen-Panel:ID095.03		
Hypertriglyzeridämie: 32 Gene (51,1 kb)		
ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEC, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24		
Hyperchylomikronämie: 6 Gene (6,1 kb)		
APOC2, APOE, APOA5, GPIHBP1, LMF1, LPL		
Lipodystrophie: 15 Gene (22,0 kb)		
ADRA2A, AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, LIPE, LMNA, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24		
Kombinierte Hyperlipidämie *	3 - 5 Wo	E
Gen-Panel: ID025.06		
Kombinierte Hyperlipidämie: 40 Gene (72,8 kb):		
ABCG5, ABCG8, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEC, CREB3L3, CYP27A1, CYP7A1, GCKR, GK, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, NPC1L1, PCSK9, PLIN1, PNPLA5, POLD1, PPARG, SORT1, ZMPSTE24		
Hypercholesterinämie: 13 Gene (36,4 kb)		
ABCG5, ABCG8, APOB, APOE, CYP7A1, CYP27A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1		
Hypertriglyzeridämie: 29 Gene (51,1 kb)		
AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEC, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PLIN1, POLD1, PPARG, ZMPSTE24		
Lipodystrophie (CGL, FPLD) *	3 - 5 Wo	E
Gen-Panel: ID055.04		
Lipodystrophie (CGL, FPLD): 15 Gene (22,0 kb)		
ADRA2A, AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, LIPE, LMNA, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24		
Kongenitale generalisierte Lipodystrophie (CGL): 5 Gene (4,9 kb)		
AGPAT2, BSCL2, CAV1, CAVIN1, PCYT1A		
Familiäre partielle Lipodystrophie (FPLD): 8 Gene (11,5 kb)		
ADRA2A, CAV1, CIDEC, LIPE, LMNA, PLAAT3, PLIN1, PPARG		
Lipodystrophien, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel: ID343.00, 45 Gene (80,0 kb)		
ADRA2A, AGPAT2, AKT2, ALDH18A1, BANF1, BSCL2, CAV1, CAVIN1, CIDEC, EPHX1, ERCC6, ERCC8, FBN1, HRAS, KCNJ6, LEP, LEPR, LIPE, LMNA, MCM3, MCM7, MFN2, MTX2, NSMCE2, OPA3, OTULIN, PCYT1A, PDGFRB, PIK3R1, PLIN1, POLD1, POLR3A, POMP, PPARG, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PTPN11, SLC25A24, SLC29A3, SPRTN, WRN, ZMPSTE24		
Statin-assoziierte Myopathie *	3 - 5 Wo	E
Gen-Panel: ID097.00, 11 Gene (37,2 kb)		
ACADM, ACADS, ACADVL, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1, SLCO1B1		

Erkrankung/Diagnostik	Dauer	Material
Gastroenterologische Erkrankungen		
Alagille-Syndrom (ALGS) *	3 - 5 Wo	E
Gen-Panel: ID112.00, 2 Gene (11,1 kb)		
JAG1, NOTCH2		
Cholestase, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID367.00, 81 Gene (179,0 kb)		
ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CCDC115, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, FAH, FOCAD, GALE, GALM, GALT, GBA1, HADHA, HNF1B, HSD3B7, IFT56, JAG1, KIF12, LARS1, LIPA, LSR, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PKHD1, POLG, RINT1, SCYL1, SEMA7A, SERPINA1, SLC25A13, SLC51A, SLC51B, SMPD1, TALDO1, TJP2, TRMU, TULP3, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, VPS50, YARS1, ZFYVE19		
Gastrointestinaler Stromatumor (GIST) *	3 - 5 Wo	E
Gen-Panel: ID226.00, 8 Gene (19,0 kb)		
KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD		
Intrahepatische Cholestase (PFIC, CBAS) *	3 - 5 Wo	E
Gen-Panel: ID159.04		
Intrahepatische Cholestase (PFIC, CBAS): 25 Gene (64,9 kb)		
ABCB4, ABCB11, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, AKR1D1, AMACR, ATP8B1, BAAT, CYP7B1, HSD3B7, JAG1, KIF12, MYO5B, NOTCH2, NR1H4, SEMA7A, SLC25A13, SLC51A, TJP2, USP53, VPS33B, ZFYVE19		
Progressive intrahepatische Cholestase (PFIC): 13 Gene (35,2 kb)		
ABCB4, ABCB11, ABCG8, ATP8B1, KIF12, MYO5B, NR1H4, SEMA7A, SLC51A, TJP2, USP53, VPS33B, ZFYVE19		
Gallensäuresynthesedefekt (CBAS): 7 Gene (10,0 kb)		
ABCD3, ACOX2, AKR1D1, AMACR, BAAT, CYP7B1, HSD3B7		
Alagille-Syndrom (ALGS): 2 Gene (11,1 kb)		
JAG1, NOTCH2		
Kolorektales Karzinom *	3 - 5 Wo	E
Gen-Panel: ID006.10, 24 Gene (66,3 kb)		
APC, ATM, AXIN2, BAP1, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53		
Lynch-Syndrom (LYNCH, HNPCC) *	2 - 4 Wo	E
Gen-Panel: ID002.02, 5 Gene (12,7 kb)		
MLH1, MSH2, MSH6, PMS2, EPCAM		
Magenkarzinom *	3 - 5 Wo	E
Gen-Panel: ID090.04, 25 Gene (73,6 kb)		
APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53		
Morbus Hirschsprung (HSCR) *	3 - 5 Wo	E
Gen-Panel: ID177.01, 20 Gene (37,1 kb)		
ECE1, EDN3, EDNRB, DNMT3B, GDNF, GFRA1, KIFBP, L1CAM, NRG1, NRTN, NTF3, NTRK3, PHOX2B, PSPN, RET, SEMA3A, SEMA3C, SEMA3D, SOX10, ZEB2		
Pankreaskarzinom *	3 - 5 Wo	E
Gen-Panel: ID089.04, 19 Gene (59,9 kb)		
APC, ATM, BARD1, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL, WT1		
Pankreatitis (PCTT) *	3 - 5 Wo	E
Gen-Panel: ID141.02, 18 Gene (29,6 kb)		
APOA5, APOC2, CASR, CEL, CFTR, CPA1, CLDN2, CELA3B, CTRC, CTSB, GPIHBP1, LMF1, LPL, PNLIP, PRSS1, SPINK1, TRPV6, UBR1		
Polyposis-Syndrom (PS, FAP) *	3 - 5 Wo	E
Gen-Panel: ID005.07, 16 Gene (41,6 kb)		
APC, AXIN2, BAP1, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11		
Polyzystische Lebererkrankung (PCLD) *	3 - 5 Wo	E
Gen-Panel: ID305.02		
Polyzystische Lebererkrankung (PCLD): 16 Gene (55,7 kb)		
ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63		
Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 7 Gene (14,0 kb)		
ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B		
Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 9 Gene (41,8 kb)		
ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1		
Viszerale Myopathien und Neuropathien, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel: ID238.02		
Viszerale Myopathien und Neuropathien, umfassende Diagnostik: 36 Gene (83,7 kb)		
ACTA2, ACTG2, CHRM3, DNMT3B, ECE1, EDN3, EDNRB, ERBB2, ERBB3, FLNA, GDNF, GFRA1, KIFBP, L1CAM, LIG3, LMOD1, MYH11, MYL9, MYLK, NRG1, NRTN, NTF3, NTRK3, PHOX2B, POLG, PSPN, RAD21, RET, RRM2B, SEMA3A, SEMA3C, SEMA3D, SGO1, SOX10, TYMP, ZEB2		
Morbus Hirschsprung (HSCR): 20 Gene (37,1 kb)		
ECE1, EDN3, EDNRB, DNMT3B, GDNF, GFRA1, KIFBP, L1CAM, NRG1, NRTN, NTF3, NTRK3, PHOX2B, PSPN, RET, SEMA3A, SEMA3C, SEMA3D, SOX10, ZEB2		
Neuropathische intestinale Pseudoobstruktion (VSCN): 9 Gene (28,6 kb)		
ERBB2, ERBB3, FLNA, LIG3, POLG, RAD21, RRM2B, SGO1, TYMP		
Myopathische intestinale Pseudoobstruktion (VSCM): 8 Gene (19,9 kb)		
ACTA2, ACTG2, CHRM3, LMOD1, MYL9, MYLK, MYH11, RAD21		

Erkrankung/Diagnostik	Dauer	Material
Gehirnfehlbildungen		
Aicardi-Goutières-Syndrom (AGS) *		
Gen-Panel: ID058.01, 9 Gene (13,0 kb)	3 - 5 Wo	E
ADAR, IFIH1, LSM11, RNASEH2A, RNASEH2B, RNASEH2C, RNU7-1, SAMHD1, TREX1		
Holoprosenzephalie (HPE) *		
Gen-Panel: ID169.02, 17 Gene (48,8 kb)	3 - 5 Wo	E
CDON, CNOT1, DLL1, DISP1, FGF8, FGFR1, GAS1, GLI2, PRRX1, PTCH1, SHH, SIX3, STAG2, STIL, TGIF1, WDR62, ZIC2		
Joubert-Syndrom (JBTS) *		
Gen-Panel: ID028.03, 40 Gene (104,1 kb)	4 - 6 Wo	E
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGAGRAM1, TTC21B, ZNF423		
Komplexe kortikale Dysplasie mit weiteren Hirnfehlbildungen (CDCBM) *		
Gen-Panel: ID271.02, 15 Gene (52,0 kb)	3 - 5 Wo	E
ADGRG1, APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2		
Lissenzephalie (LIS) *		
Gen-Panel: ID133.00, 12 Gene (46,4 kb)	3 - 5 Wo	E
ARX, CDK5, CEP85L, DCX, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A		
Meckel-Syndrom (MKS) *		
Gen-Panel: ID032.02, 13 Gene (35,1 kb)	3 - 5 Wo	E
B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231		
Neuronale Migrationsstörungen, umfassende Diagnostik *		
Gen-Panel: ID180.01	4 - 6 Wo	E
Neuronale Migrationsstörungen, umfassende Diagnostik: 82 Gene (264,4 kb)		
ACTB, ACTG1, ADGRG1, AKT3, APC2, ARF1, ARFGEF2, ARX, B3GALNT2, B4GAT1, CAMSAP1, CCND2, CDK5, CEP85L, COL3A1, COL4A1, COL4A2, COLGALT1, CRADD, CRPPA, CTNNA2, DAG1, DCHS1, DCX, DYNC1H1, EML1, EMX2, ERMARD, FAT4, FH, FIG4, FKRP, FKTN, FLNA, GMPPB, KATNB1, KIFBP, KIF2A, KIF5C, KIF26A, LAMB1, LAMC3, LARGE1, MACF1, MAP1B, MTOR, NDE1, NEDD4L, OCLN, PAFAH1B1, PHGDH, PI4KA, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSAT1, RAB18, RAB3GAP1, RAB3GAP2, RAC3, RELN, RTTN, RXYLT1, SHH, SIX3, TBC1D20, TMTC3, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2, WDR62		
Periventrikuläre noduläre Heterotopie (PVNH): 6 Gene (26,1 kb)		
ARF1, ARFGEF2, ERMARD, FLNA, MAP1B, NEDD4L		
Komplexe kortikale Dysplasie (CDCBM): 12 Gene (45,7 kb)		
APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1		
Walker-Warburg-Syndrom (MDDGA): 14 Gene (23,7 kb)		
B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1		
Lissenzephalie (LIS): 12 Gene (46,4 kb)		
ARX, CDK5, CEP85L, DCX, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A		
Polymikrogyrie: 15 Gene (42,0 kb)		
ADGRG1, AKT3, CCND2, COL3A1, FIG4, KIFBP, OCLN, PI4KA, PIK3CA, RTTN, PIK3R2, TUBA1A, TUBA8, TUBB2B, WDR62		
Schizenzephalie: 7 Gene (19,7 kb)		
COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62		
Periventrikuläre noduläre Heterotopie (PVNH) *		
Gen-Panel: ID306.00, 6 Gene (26,1 kb)	3 - 5 Wo	E
ARF1, ARFGEF2, ERMARD, FLNA, MAP1A, NEDD4L		
Polymikrogyrie *		
Gen-Panel: ID176.01, 15 Gene (42,0 kb)	3 - 5 Wo	E
ADGRG1, AKT3, CCND2, COL3A1, FIG4, KIFBP, OCLN, RTTN, PI4KA, PIK3CA, PIK3R2, TUBA1A, TUBA8, TUBB2B, WDR62		
Pontozerebelläre Hypoplasie (PCH) *		
Gen-Panel: ID071.02, 26 Gene (60,5 kb)	3 - 5 Wo	E
AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34 TSEN54, VPS51, VPS53, VRK1		
Schizenzephalie *		
Gen-Panel: ID173.00, 7 Gene (19,7 kb)	3 - 5 Wo	E
COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62		
Septooptische Dysplasie *		
Gen-Panel: ID378.00, 8 Gene (10,8 kb)	3 - 5 Wo	E
GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3		
Walker-Warburg-Syndrom (WWS, MDDGA) *		
Gen-Panel: ID178.00, 14 Gene (23,7 kb)	3 - 5 Wo	E
B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1		
Zerebelläre Dysgenesie, X-chromosomal *		
Gen-Panel: ID219.00, 11 Gene (24,5 kb)	3 - 5 Wo	E
ABCB7, CASK, DKC1, FMR1, L1CAM, MECP2, MID1, OFD1, OPHN1, SLC9A6, ZIC3		

Erkrankung/Diagnostik	Dauer	Material
Hämatologische Erkrankungen		
Anämien, umfassende Diagnostik * Gen-Panel: ID392.01 Anämien, umfassende Diagnostik: 188 Gene (357,1 kb) ABCB7, ABCG5, ABCG8, ADA, ADA2, ADAMTS13, ADH5, AK1, AK2, ALAD, ALAS2, ALDH2, ALDOA, AMN, ANK1, ATM, ATP11C, ATRX, BLM, BRCA1, BRCA2, BRIP1, C3, C1GALT1C1, CBLIF, CD46, CD59, CDAN1, CDIN1, CFB, CFH, CFHR1, CFHR3, CFI, COL4A1, COX4I2, CPOX, CUBN, CYB5R3, DGKE, DHFR, DKC1, DNASE2, DNAJC21, ELANE, EPAS1, EPB41, EPB42, EPO, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FOXP3, FTCD, FTL, G6PC3, G6PD, GATA1, GCLC, GLRX5, GPI, GPX1, GSR, GSS, HAMP, HBA1, HBA2, HBB, HBD, HBG1, HBG2, HEATR3, HFE, HK1, HSPA9, HSCB, IARS2, IREB2, KCNN4, KIF23, KLF1, LARS2, LCAT, LIG4, LPIN2, MAD2L2, MDM4, MMACHC, MMADHC, MPIG6B, MPL, MTHFD1, MTR, MTRR, MYSM1, NBN, NDUFB11, NHEJ1, NHLRC2, NHP2, NT5C3A, PALB2, PFKM, PGK1, PIEZO1, PKLR, PRF1, PUS1, RACGAP1, RAD51, RAD51C, RBSN, REN, RFWD3, RHAG, RMRP, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RUNX1, SBDS, SEC23B, SH2B3, SH2D1A, SLC11A2, SLC19A1, SLC19A2, SLC25A38, SLC30A7, SLC40A1, SLC46A1, SLC2A1, SLC4A1, SLX4, SMAD4, SPTA1, SPTB*, SRP72, STAT3, STEAP3, STIM1, TBXAS1, TCN2, TERC, TERT, TF, TFR2, THBD, THPO, TINF2, TMPRSS6, TOMM70, TP53, TPI1, TRNT1, TSR2, UMPS, UBE2T, VPS4A, WRAP53, XK, XRCC2, YARS2 Diamond-Blackfan-Anämie (DBA): 22 Gene (11,5 kb) HEATR3, RPL5, RPL9, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Sideroblastische Anämie (SIDBA): 10 Gene (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2 Megaloblastische Anämie: 13 Gene (31,4 kb) AMN, CUBN, DHFR, FTCD, MMADHC, MTHFD1, MTR, MTRR, SLC19A1, SLC19A2, SLC46A1, TCN2, UMPS Kongenitale nicht-sphärozytäre hämolytische Anämie (CNSHA): 10 Gene (15,3 kb) AK1, G6PD, GATA1, GCLC, GPI, GSR, GSS, HK1, NT5C3A, PKLR Kongenitale dyserythropoetische Anämie (CDAN): 6 Gene (12,7 kb) CDAN1, CDIN1, KIF23, KLF1, RACGAP1, SEC23B Sphärozytose (SPH) und Elliptozytose (EL): 6 Gene (26,6 kb) ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB Hereditäre Stomatozytose: 8 Gene (20,8 kb) ABCB6, ABCG5, ABCG8, KCNN4, PIEZO1, RHAG, SLC2A1, SLC4A1	4 - 6 Wo	E
Atypisches hämolytisch-urämisches Syndrom (AHUS) * Gen-Panel: ID163.04, 20 Gene (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 Wo	E
Diamond-Blackfan-Anämie (DBA) * Gen-Panel: ID394.00, 21 Gene (11,9 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL17, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2	3 - 5 Wo	E
Erythrozytose (ECYT) * Gen-Panel: ID138.02, 11 Gene (13,9 kb) BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL	3 - 5 Wo	E
Fanconi-Anämie (FANC) * Gen-Panel: ID043.02, 21 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E
Hämophilie * Gen-Panel: ID154.01, 5 Gene (18,9 kb) F8, F9, LMAN1, MCFD2, VWF	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS) * Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) * Gen-Panel: ID321.01 Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML): 121 Gene (244,1 kb): ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8 Akute myeloische Leukämie (AML): 12 Gene (28,4 kb) ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53 Diamond-Blackfan-Anämie (DBA): 20 Gene (11,3 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Shwachman-Diamond-Syndrom (SDS): 4 Gene (7,2 kb) DNAJC21, EFL1, SBDS, SRP54 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Lungenfibrose und Knochenmarkinsuffizienz (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8 Dyskeratosis congenita (DKC): 13 Gene (21,8 kb) ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53 Kongenitale Neutropenie (SCN): 10 Gene (13,9 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS Fanconi-Anämie (FANC): 20 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2 Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb) MLH1, MSH2, MSH6, PMS2	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Knochenmarkinsuffizienz (BMF) *	3 - 5 Wo	E
Gen-Panel: ID357.01		
Knochenmarkversagen (BMF): 28 Gene (47,6 kb):		
ACD, ADH5, ALDH2, DCLRE1B, DKC1, DNAJC21, DUT, EFL1, ERCC6L2, MDM4, MYSM1, NAF1, NHP2, NOP10, PARN, POT1, RPA1, RTEL1, SBDS, SLC30A7, SRP72, TERC, TERT, TINF2, TP53, TYMS, WRAP53, ZCCHC8		
Knochenmarkinsuffizienz-Syndrom (BMFS): 10 Gene (18,0 kb)		
ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, SLC30A7, SRP72, TP53		
Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 9 Gene (16,6 kb)		
NAF1, NOP10, PARN, POT1, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Dyskeratosis congenita: 12 Gene (18,2 kb)		
ACD, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53		
Neutropenie *	3 - 5 Wo	E
Gen-Panel: ID189.03		
Neutropenie: 33 Gene (55,6 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SEC61A1, SLC37A4, SMARCD2, SRP54, SRP68, TAFAZZIN, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1		
Schwere kongenitale Neutropenie (SCN): 13 Gene (19,7 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SEC61A1, SRP54, SRP68, TCIRG1, VPS45, WAS		
Syndrome mit Neutropenie: 22 Gene (38,8 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, TAFAZZIN, USB1, VPS13B, WDR1		
Porphyrie *	3 - 5 Wo	E
Gen-Panel: ID153.01, 10 Gene (12,8 kb)		
ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS		
Sideroblastische Anämie (SIDBA) *	3 - 5 Wo	E
Gen-Panel: ID355.00, 10 Gene (14,9 kb)		
ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2		
Sphärozytose (SPH) und Elliptozytose (EL) *	3 - 5 Wo	E
Gen-Panel: ID203.01		
Sphärozytose (SPH) und Elliptozytose (EL): 6 Gene (26,8 kb)		
ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB		
Sphärozytose (SPH): 5 Gene (24,2 kb)		
ANK1, EPB42, SLC4A1, SPTA1, SPTB		
Elliptozytose (EL): 4 Gene (18,4 kb)		
EPB41, SLC4A1, SPTA1, SPTB		
Thrombophilie (THPH) *	3 - 5 Wo	E
Gen-Panel: ID150.01, 12 Gene (25,2 kb)		
F2, F5, F9, F13B, HABP2, HRG, MTHFR, PROC, PROS1, SERPINC1, SERPIND1, THBD		
Thrombotische Mikroangiopathie (TMA) *	3 - 5 Wo	E
Gen-Panel: ID707.00, 23 Gene (44,7 kb)		
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD		
Thrombozytendefekte, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID274.02		
Thrombozytendefekte, umfassende Diagnostik: 72 Gene (155,0 kb)		
ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, EPHB2, ETV6, FERMT3, FLI1, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF1, IKZF5, ITGA2, ITGA2B, ITGB3, JAK2, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, PTPRJ, RAP1B, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TPM4, TUBA8, TUBB1, WAS, WDR1, WIPF1		
Thrombozytopenie, nicht-syndromal (THC): 16 Gene (29,1 kb)		
ACTB, ANKRD26, CYCS, ETV6, FYB1, GALE, GNE, IKZF5, MASTL, PTPRJ, RAP1B, SRC, THPO, TUBB1, TUBA8, WAS		
Thrombozythämie (THCYT): 3 Gene (6,4 kb)		
JAK2, MPL, THPO		
Thrombozytenbedingte Blutungsstörung (BDPLT): 24 Gene (53,6 kb)		
ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA*, GP1BB, GP6, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1, TPM4		
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Thrombozytopathie (BDPLT, HPS) *	3 - 5 Wo	E
Gen-Panel: ID119.01		
Thrombozytopathie (BDPLT, HPS): 37 Gene (91,0 kb)		
ACTN1, ANO6, AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CD36, DTNBP1, EPHB2, FERMT3, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1		
Thrombozytenbedingte Blutungsstörung (BDPLT) mit Thrombozytopenie: 15 Gene (37,9 kb)		
ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14		
Thrombozytenbedingte Blutungsstörung (BDPLT) ohne Thrombozytopenie: 10 Gene (20,3 kb)		
ANO6, EPHB2, GP6, ITGA2B, ITGB3, P2RY12, PTGS1, RASGRP2, TBXA2R, TBXAS1		
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Thrombozytopenie *	3 - 5 Wo	E
Gen-Panel: ID104.03		
Thrombozytopenie (THC): 49 Gene (98,5 kb)		
ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CD36, CDC42, CYCS, DIAPH1, ETV6, FLI1, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP9, HOXA11, IKZF1, IKZF5, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, PLAU, PRKACG, PTPRJ, RAP1B, RBM8A, RUNX1, SLFN14, SRC, STIM1, THPO, TPM4, TUBA8, TUBB1, WAS*, WDR1, WIPF1		
Thrombozytopenie, nicht-syndromal (THC): 13 Gene (26,3 kb)		
ANKRD26, CYCS, ETV6, FYB1, GNE, IKZF5, MASTL, PTPRJ, SRC, THPO, TUBA8, TUBB1, WAS		
Thrombozytenbedingte Blutungsstörung (BDPLT): 16 Gene (38,8 kb)		
ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14, TPM4		

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Adams-Oliver-Syndrom (AOS) *	3 - 5 Wo	E
Gen-Panel: ID259.00, 6 Gene (23,2 kb)		
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		
Albinismus, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel: ID175.05		
Albinismus, umfassende Diagnostik: 33 Gene (73,8 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1		
Okulokutaner Albinismus (OCA, OA): 9 Gene (13,2 kb)		
DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Waardenburg-Syndrom (WS): 8 Gene (9,3 kb)		
EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR		
Griscelli-Syndrom (GS): 3 Gene (8,0 kb)		
MLPH, MYO5A, RAB27A		
Amelogenesis imperfecta (AI) *	3 - 5 Wo	E
Gen-Panel: ID232.01, 17 Gene (29,4 kb)		
ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAPH, RELT, SLC24A4, WDR72		
Cowden-Syndrom (CWS) *	3 - 5 Wo	E
Gen-Panel: ID075.01, 8 Gene (12,8 kb)		
AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1		
Cutis laxa (ARCL, ADCL) *	3 - 5 Wo	E
Gen-Panel: ID109.03, 13 Gene (27,9 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1, RIN2		
Dyskeratosis congenita (DKC) *	3 - 5 Wo	E
Gen-Panel: ID347.01, 15 Gene (24,1 kb)		
ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53		
Ektodermale Dysplasie (ECTD) *	3 - 5 Wo	E
Gen-Panel: ID136.04		
Ektodermale Dysplasie (ECTD): 44 Gene (74,6 kb)		
AP1B1, CDH3, CHUK, CST6, DLX3, DSG4, EDA, EDAR, EDARADD, GJA1, GJB2, GJB6, GRHL2, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LRP6, MSX1, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, WNT10A		
Ektodermale Dysplasie (ECTD), nicht-syndromal: 20 Gene (27,5 kb)		
CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A		
Ektodermale Dysplasie (ECTD), syndromal: 28 Gene (51,0 kb)		
AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2		
Ektodermale Dysplasien, umfassende Diagnostik	4 - 6 Wo	E
Gen-Panel: ID366.00		
Ektodermale Dysplasien, umfassende Diagnostik: 92 Gene (206,6 kb)		
ANAPC1, ANTXR1, AP1B1, APCDD1, ARID1A, ARID1B ATP6V1B2, AXIN2, C3ORF52, CDH1, CDH3, CDSN, CHUK, CST6, CTNND1, CTSK, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, EVC, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB2, GJB6, GRHL2, HEPHL1, HOXC13, HR, IFT122, IFT140, IFT43, IFT52, IKBKG, INSR, KCTD1, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LIPH, LPAR6, LRP6, MBTPS2, MSX1, NECTIN1, NECTIN4, NFKB2, NFKBIA, NLRP1, PAX9, PEX1, PEX6, PKP1, PORCN, PRKD1, RIPK4, ROGDI, RSPO4, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SNRPE, SREBF1, ST14, TBC1D24, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, UBR1, WDR19, WDR35, WNT10A		
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), nicht-syndromal: 20 Gene (27,5 kb)		
CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A		
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), syndromal: 28 Gene (51,0 kb)		
AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2		
Komplexe Syndrome mit ektodermaler Dysplasie: 42 Gene (120,5 kb)		
ANTXR1, ARID1A, ARID1B, ATP6V1B2, CDH1, CTNND1, CTSK, DSG4, DSP, EVC, EVC2, FGF10, FGFR2, FGFR3, HEPHL1, IFT122, IFT140, IFT43, IFT52, INSR, KCTD1, KRT14, KRT16, KRT17, KRT81, KRT83, KRT86, NLRP1, PEX1, PEX6, ROGDI, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SREBF1, TBC1D24, UBR1, WDR19, WDR35		
Epidermolysis bullosa (EB) *	4 - 6 Wo	E
Gen-Panel: ID198.02		
Epidermolysis bullosa (EB): 34 Gene (120,6 kb)		
ATP2A2, ATP2C1, CAST, CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FERMT1, FLG2, IKBKG, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5		
Epidermolysis bullosa simplex (EBS): 7 Gene (33,4 kb)		
CD151, DST, EXPH5, KLHL24, KRT14, KRT5, PLEC		
Epidermolysis bullosa junctionalis (JEB): 7 Gene (28,4 kb)		
COL17A1, ITGA3, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2		
Epidermolysis bullosa dystrophica (DEB): 1 Gen (8,8 kb)		
COL7A1		
Kindler Epidermolysis bullosa (KNDLRS): 1 Gen (2,0 kb)		
FERMT1		
Syndrome mit Epidermolysis bullosa: 20 Gene (63,9 kb)		
ATP2A2, ATP2C1, CAST, CD151, CDSN, CHST8, CSTA, DSG1, DSP, FERMT1, FLG2, IKBKG, ITGA3, JUP, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5		
Gorlin-Syndrom (BCNS) und ähnliche Krankheitsbilder *	3 - 5 Wo	E
Gen-Panel: ID174.02, 8 Gene (25,8 kb)		
BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU		

Erkrankung/Diagnostik	Dauer	Material
Hereditäres Angioödem (HAE) *	3 - 5 Wo	E
Gen-Panel: ID345.00, 7 Gene (16,4 kb)		
ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1		
Hermansky-Pudlak-Syndrom (HPS) *	3 - 5 Wo	E
Gen-Panel: ID289.00, 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Hydrops fetalis *	4 - 6 Wo	E
Gen-Panel: ID370.00, 148 Gen (404,7 kb)		
ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNG, COL2A1, CTSA, DHCR24, DHCR7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHBP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUFB10, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148		
Hypotrichose, nicht-syndromale Form (HYPT) *	3 - 5 Wo	E
Gen-Panel: ID146.01, 12 Gene (20,1 kb)		
APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE		
Ichthyose *	3 - 5 Wo	E
Gen-Panel: ID217.01		
Ichthyose: 38 Gene (73,0 kb)		
ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CLDN10, CSTA, CYP4F22, ELOVL4, ERCC2, FLG, GJB2, KRT1, KRT10, KRT2, LIPN, LORICRIN, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, TGM1		
Kongenitale Ichthyose, autosomal-rezessiv (ARCI): 14 Gene (28,7 kb)		
ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1		
Ichthyose, autosomal-dominant und X-chromosomal: 7 Gene (21,3 kb)		
ASPRV1, FLG, GJB2, KRT1, KRT2, KRT10, STS		
Kutanes malignes Melanom (CMM) *	3 - 5 Wo	E
Gen-Panel: ID193.01, 12 Gene (26,4 kb)		
BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3		
Multiples Pterygium-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID158.01, 8 Gene (16,8 kb)		
CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4		
Neurofibromatose (NF) *	3 - 5 Wo	E
Gen-Panel: ID210.00, 3 Gene (11,6 kb)		
NF1, NF2, SPRED1		
Okulokutaner Albinismus (OCA) *	3 - 5 Wo	E
Gen-Panel: ID082.02, 9 Gene (13,3 kb)		
DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Orofaziodigitales Syndrom (OFD) *	3 - 5 Wo	E
Gen-Panel: ID265.01, 14 Gene (40,2 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TBC1D32, TCTN3, TMEM107, ZRSR2		
Pachyonychia congenita (PC) *	3 - 5 Wo	E
Gen-Panel: ID120.01, 13 Gene (30,1 kb)		
AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3		
Palmoplantarkeratose (PPK) *	3 - 5 Wo	E
Gen-Panel: ID323.00		
Palmoplantarkeratose (PPK): 33 Gene (58,2 kb)		
AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB3, GJB4, JUP, KDSR, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT10, KRT14, KRT16, KRT17, KRT83, LORICRIN, MBTPS2, PERP, RHBDF2, SERPINB7, SLURP1, SMARCAD1, TAT, TRPM4, TRPV3, WNT10A		
Palmoplantarkeratose (PPK), nicht-syndromal: 11 Gene (25,7 kb)		
AAGAB, AQP5, DSG1, DSP, KRT1, KRT6C, KRT9, KRT10, KRT16, SERPINB7, TRPV3		
Palmoplantarkeratose (PPK), syndromal: 15 Gene (22,4 kb)		
CTSC, ENPP1, GJA1*, GJB2, KRT14, MBTPS2, JUP, LORICRIN, PERP, RHBDF2, SLURP1, SMARCAD1, TAT, TRPV3, WNT10A		
Erythrokeratodermia variabilis et progressiva (EKVP): 7 Gene (9,5 kb)		
GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4		
Pachyonychia congenita (PC): 4 Gene (6,1 kb)		
KRT6A, KRT6B, KRT16, KRT17		
Peeling-Skin-Syndrom (PSS) *	3 - 5 Wo	E
Gen-Panel: ID309.00, 6 Gene (13,6 kb)		
CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5		
Pierre-Robin-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID294.01, 34 Gene (73,7 kb)		
AMER1, ARCN1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, GNAI3, MYMK, MYMX, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5		
Primäres Lymphödem (LMPHM) *	3 - 5 Wo	E
Gen-Panel: ID372.00, 21 Gene (69,4 kb)		
ADAMTS3, ANGPT2, CALCRL, CCBE1, CELSR1, EPHB4, ERG, FAT4, FLT4, FOXC2, GATA2, GJA1, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, THSD1, TIE1, VEGFC		
Progerie und progeroide Syndrome *	3 - 5 Wo	E
Gen-Panel: ID147.01, 25 Gene (61,9 kb)		
ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24		
Selektive Zahn-Agenesie (STHAG) *	3 - 5 Wo	E
Gen-Panel: ID151.02, 16 Gene (26,9 kb)		
EDA, EDAR, EDARADD, GREM2, IRF6, KDF1, KREMEN1, LRP6, LTBP3, MSX1, PAX9, PTH1R, TP63, TSPEAR, WNT10A, WNT10B		
Tuberöse Sklerose (TSC) *	2 - 4 Wo	E
Gen-Panel: ID332.00, 2 Gene (8,9 kb)		
TSC1, TSC2		

Erkrankung/Diagnostik	Dauer	Material
Systemerkrankungen mit multiplen Café-au-lait-Flecken *	3 - 5 Wo	E
Gen-Panel ID351.00		
Systemerkrankungen mit multiplen Café-au-lait-Flecken: 9 Gene (27,6 kb)		
BRAF, MLH1, MSH2, MSH6, NF1, PMS2, PTPN11, RAF1, SPRED1		
Konstitutionelle Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb)		
MLH1, MSH2, MSH6, PMS2		
Neurofibromatose (NF1, NFLS): 2 Gene (9,8 kb)		
NF1, SPRED1		
LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb)		
BRAF, PTPN11, RAF1		
Xeroderma pigmentosum (XP) *	3 - 5 Wo	E
Gen-Panel: ID282.00, 10 Gene (23,5 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC		
Herz- und Gefäßerkrankungen		
Alagille-Syndrom (ALGS) *	3 - 5 Wo	E
Gen-Panel: ID112.00, 2 Gene (11,1 kb)		
JAG1, NOTCH2		
Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC) *	4 - 6 Wo	E
Gen-Panel: ID010.03, 27 Gene (185,3 kb)		
ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN		
Atriumseptumdefekt und Ventrikelseptumdefekt (ASD, VSD, AVSD) *	3 - 5 Wo	E
Gen-Panel: ID143.00, 12 Gene (21,1 kb)		
ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1		
Bikuspide Aortenklappe (AOVD) *	3 - 5 Wo	E
Gen-Panel: ID301.00, 6 Gene (16,7 kb)		
GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID137.06		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik: 85 Gene (283,3 kb)		
ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ASPH, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, DCC, DLG4, DSE, EFEMP1, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, IPO8, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NKAP, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THBS2, THSD4, TNXB, ZNF469		
Marfan-Syndrom (MFS): 3 Gene (11,8 kb)		
FBN1, TGFBR2, TGFBR1		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 18 Gene (45,7 kb)		
ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Cutis laxa-Syndrom (ARCL, ADCL): 12 Gene (29,4 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
Stickler-Syndrom (STL): 6 Gene (22,0 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2		
Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Brugada-Syndrom (BRGDA) *	3 - 5 Wo	E
Gen-Panel: ID014.02, 23 Gene (56,4 kb)		
ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4		
CADASIL und CARASIL *	3 - 5 Wo	E
Gen-Panel: ID167.01, 3 Gene (9,4 kb)		
HTRA1, NOTCH3, TREX1		
CHARGE-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID307.00, 3 Gene (12,9 kb)		
CHD7, SEMA3E, TBX22		
Cutis laxa (ARCL, ADCL) *	3 - 5 Wo	E
Gen-Panel: ID109.03, 13 Gene (27,9 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1, RIN2		
Dilatative Kardiomyopathie (CMD, DCM) *	4 - 6 Wo	E
Gen-Panel: ID008.05, 68 Gene (301,9 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1		
Ehlers-Danlos-Syndrom (EDS) *	3 - 5 Wo	E
Gen-Panel: ID039.06		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 9 Gene (39,6 kb)		
C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1, THBS2		
Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469		

Erkrankung/Diagnostik	Dauer	Material
Endokrine Hypertonie * Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E
Fallot-Tetralogie (TOF) * Gen-Panel: ID144.01, 12 Gene (23,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2	3 - 5 Wo	E
Frühes Repolarisationssyndrom (ERS) * Gen-Panel: ID330.00, 12 Gene (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A	3 - 5 Wo	E
Herz-Hand-Syndrom * Gen-Panel: ID165.01, 9 Gene (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 Wo	E
Hypertrophe Kardiomyopathie (CMH, HCM) * Gen-Panel: ID007.05, 56 Gene (253,8 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, LAMP2, KRAS, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL	4 - 6 Wo	E
Isolierte kongenitale Herzfehler * Gen-Panel: ID017.04 Isolierte kongenitale Herzfehler: 43 Gene (103,7 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Atrium-/Ventrikelseptumdefekt (ASD, VSD): 12 Gene (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NR2F2, NKX2-5, TBX20, TLL1 Fallot-Tetralogie (TOF): 12 Gene (23,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2 Viszerale Heterotaxie (HTX): 13 Gene (26,2 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, MMP21, MNS1, NODAL, PKD1L1, ZIC3 Konotrunkale Herzfehlbildung (CTHM): 12 Gene 24,8 (kb) FLT4, FOXH1, CFC1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3 Aortenklappenerkrankung (AOVD): 7 Gene (19,1 kb) ELN, GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2 Hypoplastisches Linksherzsyndrom (HLHS): 5 Gene (5,2 kb) GDF1, GJA1, HAND1, NKX2-5, NR2F2 Kongenitale multiple Herzfehlbildung (CHTD): 7 Gene (14,4 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, TAB2, ZIC3 Herzklappendysplasie (CVDP): 3 Gene (14,8 kb) ADAMTS19, FLNA, PLD1	4 - 6 Wo	E
Kongenitale Herzfehler, umfassende Diagnostik * Gen-Panel: ID019.02 Kongenitale Herzfehler, umfassende Diagnostik: 149 Gene (472,9 kb) ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR2B, ADAMTS10, ADAMTS17, ADAMTS19, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CFAP45, CFAP52, CFAP53, CFC1, CHD4, CHD7, CIROP, CITED2, CREBBP, CRELD1, DHCR7, DLL4, DNAAF1, DNAH5, DNAH9, DNAH11, DOCK6, DPYSL5, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FLT4, FOXC1, FOXF1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAAO, HAND1, HOXA1, HRAS, ISL1, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MMP21, MNS1, MRAS, MYH11, MYH6, MYRF, NADSYN1, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2, NR2F2, NRAS, NSD1, ODAD2, PIGL, PITX2, PKD1L1, PLD1, PPP1CB, PRDM6, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, ROBO4, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMAD6, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, SPRED2, STAG2, STRA6, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TKT, TLL1, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZFPM2, ZIC3 Isolierte kongenitale Herzfehler: 48 Gene (149,9 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Syndromale kongenitale Herzfehler: 109 Gene (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Kardiale Arrhythmien, umfassende Diagnostik * Gen-Panel: ID026.03 Kardiale Arrhythmien, umfassende Diagnostik: 71 Gene (274,2 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, EMD, FGF12, GJA5, GNB2, GNB5, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYH6, MYL4, NKX2-5, NPPA, NUP155, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TANGO2, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN Long-QT-Syndrom (LQT): 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN Brugada-Syndrom (BRGDA): 23 Gene (56,4 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPDL, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2V, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4 Frühes Repolarisationssyndrom (ERS): 12 Gene (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A Familiäres Vorhofflimmern (ATFB): 17 Gene (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A Short-QT-Syndrom (SQT): 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCL4A3 Sick-Sinus-Syndrom (SSS): 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN Arrhythmogene rechtsventrikuläre Dysplasie (ARVD): 15 Gene (150,6 kb) CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, PLN, LMNA, PKP2, PRKAG2, RYR2, TGFB3, TMEM43, TTN	4 - 6 Wo	E
Kardiomyopathien, umfassende Diagnostik * Gen-Panel: ID027.05 Kardiomyopathien, umfassende Diagnostik: 154 Gene (487,0 kb) ABCC9, ACTA1, ACTC1, ACTN2, ALPK3, ANKRD1, APOA1, B2M, BAG3, BAG5, BRAF, CACNA1C, CACNB2, CALR3, CAP2, CASQ2, CAV3, CDH2, COA5, COA6, CORIN, COX15, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DMPK, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGA, FHL1, FHL2, FHOD3, FKRP, FKTN, FLII, FLNC, FTH1, FXN, GAA, GATA4, GATAD1, GET3, GLA, GSN, HADHA, HAMP, HCN4, HFE, HJV, HRAS, ILK, JPH2, JUP, KCNQ1, KIF20A, KLF10, KLHL24, KRAS, KY, LAMA4, LAMP2, LDB3, LIMS2, LMNA, LMOD2, LRRC10, LYZ, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM10, MIB1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOT, MYOZ2, MYPN*, NEBL, NEXN, NKX2-5, NNT, NONO, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, PYROXD1, RAF1, RBM20, RIT1, RPL3L, RRAS2, RYR2, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC40A1, SLC25A4, SOS1, SOS2, SPRED2, SVIL, SYNE1, SYNE2, TAFAZZIN, TBX5, TBX20, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TRPM4, TTN, TTR, UNC45B, VCL, VEZF1 Dilatative Kardiomyopathie (DCM, CMD): 68 Gene (301,9 kb) ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1 Hypertrophe Kardiomyopathie (HCM, CMH): 56 Gene (253,8 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL Restriktive Kardiomyopathie (RCM): 15 Gene (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC): 27 Gene (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN Nichtdilatierte linksventrikuläre Kardiomyopathie (NDLVC, LVNC): 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) * Gen-Panel: ID012.03, 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN	3 - 5 Wo	E
Konotrunkale Herzfehlbildung (CTHM) * Gen-Panel: ID160.02, 18 Gene (42,8 kb) FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, JAG1, NKX2-5, NKX2-6, NOTCH1, NR2F2, SMAD2, PLXND1, TAB2, TBX1, TMEM260, ZFPM2, ZIC3	3 - 5 Wo	E
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.08 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 40 Gene (117,6 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (12,6 kb) BGN, IPO8, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit thorakalem Aortenaneurysma: 31 Gene (96,5 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, IPO8, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E
Long-QT-Syndrom (LQT) * Gen-Panel: ID013.01, 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN	3 - 5 Wo	E
Marfan-Syndrom (MFS) * Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1 TGFBR1, TGFBR2	2 - 4 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.06 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 50 Gene (157,9 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP1, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 17 Gene (42,1 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Stickler-Syndrom (STL): 6 Gene (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Syndrome mit marfanoidem Habitus: 20 Gene (52,2 kb) CBS, DLG4, EFEMP1, EFEMP2, FBN1, FBN2, NKAP, NPR2, MED12, PLOD1, PRDM5, SKI, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZDHHC9	4 - 6 Wo	E
Multipler kongenitaler Herzdefekt (CHTD) * Gen-Panel: ID302.01, 9 Gene (21,5 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, PLXND1, SMAD2, TAB2, ZIC3	3 - 5 Wo	E
Muskelerkrankungen mit Herzbeteiligung * Gen-Panel: ID123.03 Muskelerkrankungen mit Herzbeteiligung: 19 Gene (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss-Muskeldystrophie (EMDM): 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker-Muskeldystrophie (DMD, BMD): 1 Gen (11,1 kb) DMD	4 - 6 Wo	E
Nichtdilatierte linksventrikuläre Kardiomyopathie (NDLVC, LVNC) * Gen-Panel: ID011.04, 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
Noonan-Syndrom (NS) * Gen-Panel: ID023.06, 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 Wo	E
Plötzlicher Herztod * Gen-Panel: ID349.01 Plötzlicher Herztod: 127 Gene (393,9 kb) ABCC9, ACTC1, ACTN2, AKAP9, ALG10B, ALPK3, ANK2, ANKRD1, BAG3, BAG5, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAP2, CASQ2, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGF12, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GJA5, GLA, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLHL24, LAMA4, LAMP2, LDB3, LEMD2, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, NPPA, NUP155, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RANGRF, RBM20, RPL3L, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SEMA3A, SGCD, SLC4A3, SLMAP, SNTA1, TAFAZZIN, TBX5, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSPYL1, TTN, TTR, VCL, VEZF1 Kardiale Arrhythmien (BRGDA, LQT) und plötzlicher Herztod: 74 Gene (281,6 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FGF12, GJA5, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYBPC3, MYH6, MYL4, NKX2-5, NPPA, NUP155, PPA2, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TSPYL1, TTN Kardiomyopathien (HCM, DCM) und plötzlicher Herztod: 71 Gene (261,3 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, BAG5, CAP2, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSG2, DSP, EMD, EYA4, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GLA, JPH2, JUP, KLHL24, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, TAFAZZIN, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL, VEZF1	4 - 6 Wo	E
Pulmonale Hypertonie (PAH) * Gen-Panel: ID281.01, 23 Gene (49,9 kb) ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4	3 - 5 Wo	E
RASopathien * Gen-Panel: ID015.05 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan-Syndrom (NS): 15 Gene (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 Wo	E
Restriktive Kardiomyopathie (RCM) * Gen-Panel: ID105.03, 15 Gene (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR	4 - 6 Wo	E
Short-QT-Syndrom (SQT) * Gen-Panel: ID233.01, 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3	3 - 5 Wo	E
Sick-Sinus-Syndrom (SSS) * Gen-Panel: ID107.01, 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Speicherkrankheiten mit Herzbeteiligung * Gen-Panel: ID149.03 Speicherkrankheiten mit Herzbeteiligung: 16 Gene (20,7 kb) APOA1, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Kardiale Glykogenspeicherkrankheit (GSD): 3 Gene (5,8 kb) GAA, LAMP2, PRKAG2 Hämochromatose (HFE): 6 Gene (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Amyloidose (AMYLD): 6 Gene (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR	3 - 5 Wo	E
Syndromale kongenitale Herzfehler * Gen-Panel ID252.02 Syndromale kongenitale Herzfehler: 109 Gene (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3 Noonan-Syndrom (NS): 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 Ritscher-Schinzel-Syndrom (RTSC): 4 Gene (10,0 kb) DPYSL5, CCDC22, VPS35L, WASHC5 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Adams-Oliver-Syndrom (AOS): 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ Kabuki-Syndrom (KABUK): 2 Gene (20,8 kb) KDM6A, KMT2D Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 CHARGE-Syndrom: 2 Gene (11,3 kb) CHD7, SEMA3E VCRL-Syndrom: 3 Gene (4,4 kb) HAAO, KYNU, NADSYN1	4 - 6 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (TAA/D) * Gen-Panel: ID020.02 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 17 Gene (44,1 kb) ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	3 - 5 Wo	E
Viszerale Heterotaxie (HTX) * Gen-Panel: ID145.01, 18 Gene (72,4 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, DNAAF1, DNAH5, DNAH9, DNAH11, MMP21, MNS1, NODAL, ODAD2, PKD1L1, ZIC3	3 - 5 Wo	E
Vorhofflimmern (ATFB) * Gen-Panel: ID016.02, 17 Gene (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A	3 - 5 Wo	E
Zerebrale Kleingefäßerkrankung (BSVD) * Gen-Panel: ID325.00, 3 Gene (12,0 kb) COL4A1, COL4A2, COLGALT1	3 - 5 Wo	E
Zerebrovaskuläre Erkrankungen und Schlaganfall * Gen-Panel: ID234.02 Zerebrovaskuläre Erkrankungen und Schlaganfall: 44 Gene (130,0 kb) ACE, ADA2, ACTA2, ALOX5AP, APOE, APP, CBS, CCM2, COL3A1, COL4A1, COL4A2, COLGALT1, CST3, F2, F5, FBN1, FLNA, GAA, GLA, GSN, GUCY1A1, HTRA1, ITM2B, JAG1, KRIT1, MTHFR, MYH11, MYLK, NOS3, NOTCH3, OTC, PDCD10, POLG, PRKCH, PRNP, RNF213, SLC2A10, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, TREX1, TTR Zerebrale Amyloidangiopathie: 6 Gene (7,1 kb) APP, CST3, GSN, ITM2B, PRNP, TTR Zerebrale Kleingefäßerkrankung (BSVD): 3 Gene (12,0 kb) COL4A1, COL4A2, COLGALT1 Zerebrale kavernöse Fehlbildung (CCM): 3 Gene (4,2 kb) CCM2, KRIT1, PDCD10 Zerebrales Aneurysma und Dissektion: 10 Gene (32,8 kb) ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Moyamoya-Erkrankung (MYMY): 4 Gene (22,2 kb) ACTA2, GUCY1A1, JAG1, RNF213 CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
HNO-Erkrankungen		
Branchiootorenales Syndrom (BOR) * Gen-Panel: ID315.00, 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 Wo	E
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) * Gen-Panel: ID170.05, 40 Gene (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 Wo	E
Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA) * Gen-Panel: ID091.04, 60 Gene (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1	4 - 6 Wo	E
Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB) * Gen-Panel: ID092.04, 83 Gene (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C*, WBP2, WHRN	4 - 6 Wo	E
Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik * Gen-Panel: ID237.03 Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik: 133 Gene (374,8 kb) ABCC1, ACTG1, ADCY1, AFG2B, AIFM1, ATOH1, ATP11A, ATP2B2, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GPRASP2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MAP1B, MARVELD2, MCM2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PI4KB, PJVK, PKHD1L1, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, THOC1, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, USP48, WBP2, WFS1, WHRN Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA): 60 Gene (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24*, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1 Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB): 83 Gene (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX): 6 Gene (11,8 kb) AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX	4 - 6 Wo	E
Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX) * Gen-Panel: ID290.01, 8 Gene (17,1 kb) AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX, TIMM8A	3 - 5 Wo	E
Perrault-Syndrom (PRLTS) * Gen-Panel: ID388.00, 9 Gene (14,1 kb) CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, MRPL49, PRORP, TWNK	3 - 5 Wo	E
Syndromale Schwerhörigkeit, umfassende Diagnostik * Gen-Panel: ID190.01 Syndromale Schwerhörigkeit, umfassende Diagnostik: 109 Gene (317,3 kb) ABHD12, ADGRV1, AIFM1, ALMS1, ANKH, ARSG, ATP1A3, ATP6V0A4, ATP6V1B1, BCAP31, BCS1L, BRAF, BSND, CACNA1D, CATSPER2, CD151, CDH23, CHD7, CHSY1, CIB2, CISD2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, DCAF17, DIAPH3, DLX5, DNMT1, DSPP, EDN3, EDNRB, ERAL1, ESPN, EXOSC2, EYA1, FGF3, FGFR3, FOXC1, FOXI1, GATA3, GJA1, GJB2, GPSM2, HARS1, HARS2, HSD17B4, KCNE1, KCNJ10, KCNQ1, KITLG, LARS2, LHX3, LRP2, MAF, MANBA, MITF, MPZ, MYH14, MYH9, MYO7A, NLRP3, PAX3, PCDH15, PDZD7, PEX1, PEX6, PLOD3, PMP22, POLD1, POLR1C, POLR1D, PRDM5, PRPS1, PTPN11, RAF1, RPGR, RPS6KA3, SALL1, SALL4, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLC4A11, SLC52A2, SLC52A3, SNAI2, SOX10, SPATA5, TCOF1, TFAP2A, TIMM8A, TWNK, TYR, USH1C, USH1G, USH2A, WFS1, WHRN, ZNF469 Usher-Syndrom (USH): 13 Gene (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USHC, USH1G, USH2A, WHRN Stickler-Syndrom (STL): 6 Gene (21,2 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Alport-Syndrom (ATS): 4 Gene (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9 Waardenburg-Syndrom (WS): 8 Gene (9,4 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Perrault-Syndrom (PRLTS): 6 Gene (9,3 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1 CHARGE-Syndrom: 2 Gene (11,3 kb) CHD7, SEMA3E	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Usher-Syndrom (USH) *		
Gen-Panel: ID034.01		
Usher-Syndrom (USH): 13 Gene (70,3 kb)		
ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN		
Usher-Syndrom, Typ I (USH1): 6 Gene (26,2 kb)		
CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G		
Usher-Syndrom, Typ II (USH2): 4 Gene (40,4 kb)		
ADGRV1, PDZD7, USH2A, WHRN		
Usher-Syndrom, Typ III und Typ IV (USH3, USH4): 3 Gene (3,8 kb)		
ARSG, CLRN1, HARS1	3 - 5 Wo	E
Immunologische Erkrankungen		
Antikörpermangelsyndrome (CVID, AGM) *		
Gen-Panel: ID382.00		
Antikörpermangelsyndrome (CVID, AGM): 38 Gene (70,7 kb)		
AICDA, BLNK, BTK, CARD11, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, CTNNBL1, CXCR4, FNIP1, ICOS, IGLL1, IKZF1, IL21, IRF2BP2, LRBA, LRRC8A, MS4A1, NFKB1, NFKB2, OAS1, PIK3CD, PIK3R1, PLCG2, RAC2, SEC61A1, SH3KBP1, SLC39A7, SPI1, TCF3, TNFRSF13B, TNFRSF13C, TOP2B, UNG		
Allgemeiner variabler Immundefekt (CVID): 14 Gene (28,0 kb)		
CD19, CD81, CR2, ICOS, IKZF1, IL21, IRF2BP2, LRBA, MS4A1, NFKB1, NFKB2, SEC61A1, TNFRSF13B, TNFRSF13C		
Agammaglobulinämie (AGM): 11 Gene (16,2 kb)		
BLNK, BTK, CD79A, CD79B, IGLL1, LRRC8A, PIK3R1, SH3KBP1, SLC39A7, SPI1, TCF3		
Immundefekt mit Hyper-IgM (HIGM): 4 Gene (3,2 kb)		
AICDA, CD40, CD40LG, UNG	3 - 5 Wo	E
Atypisches hämolytisch-urämisches Syndrom (AHUS) *		
Gen-Panel: ID163.04, 20 Gene (42,1 kb)		
ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 Wo	E
Autoinflammatorische Erkrankungen, umfassende Diagnostik *		
Gen-Panel: ID087.05		
Autoinflammatorische Syndrome, umfassende Diagnostik: 51 Gene (99,3 kb)		
ADA2, ALPK1, ARPC1B, ARPC5, CARD14, DNASE2, DOCK11, DPP9, ELANE, ELF4, HCK, IKBKG, IL1RN, IL36RN, JAK1, LPIN2, LYN, MEFV, MVK, NCKAP1L, NFKB1, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NTRK1, OTULIN, PLCG2, POMP, PSMB4, PSMB8, PSMB9, PSMB10, PSMG2, PSTPIP1, RBCK1, RELA, RIPK1, RNF31, SHARPIN, SLC29A3, SOCS1, STING1, SYK, TBK1, TNFAIP3, TNFRSF1A, TNFRSF11A, TRNT1, WDR1		
Periodische Fiebersyndrome: 14 Gene (29,9 kb)		
ELANE, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, PSMB8, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 Wo	E
Chronische Granulomatose (CGD) *		
Gen-Panel: ID379.00, 8 Gene (8,8 kb)		
CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2	3 - 5 Wo	E
Hydrops fetalis *		
Gen-Panel: ID370.00, 148 Gen (404,7 kb)		
ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNG, COL2A1, CTSA, DHCR24, DHCR7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHBP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUFB10, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	4 - 6 Wo	E
Hyper-IgE-Syndrom mit rekurrenten Infektionen (HIES) *		
Gen-Panel: ID240.01, 10 Gene (29,5 kb)		
DOCK8, DSG1, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TYK2, ZNF341	3 - 5 Wo	E
Knochenmarkinsuffizienz (BMF) *		
Gen-Panel: ID357.01		
Knochenmarkversagen (BMF): 28 Gene (47,6 kb):		
ACD, ADH5, ALDH2, DCLRE1B, DKC1, DNAJC21, DUT, EFL1, ERCC6L2, MDM4, MYSM1, NAF1, NHP2, NOP10, PARN, POT1, RPA1, RTEL1, SBDS, SLC30A7, SRP72, TERC, TERT, TINF2, TP53, TYMS, WRAP53, ZCCHC8		
Knochenmarkinsuffizienz-Syndrom (BMFS): 10 Gene (18,0 kb)		
ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, SLC30A7, SRP72, TP53		
Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 9 Gene (16,6 kb)		
NAF1, NOP10, PARN, POT1, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Dyskeratosis congenita: 12 Gene (18,2 kb)		
ACD, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53	3 - 5 Wo	E
Neutropenie *		
Gen-Panel: ID189.03		
Neutropenie: 33 Gene (55,6 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SEC61A1, SLC37A4, SMARCD2, SRP54, SRP68, TAFAZZIN, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1		
Schwere kongenitale Neutropenie (SCN): 13 Gene (19,7 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SEC61A1, SRP54, SRP68, TCIRG1, VPS45, WAS		
Syndrome mit Neutropenie: 22 Gene (38,8 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, TAFAZZIN, USB1, VPS13B, WDR1	3 - 5 Wo	E
Periodische Fiebersyndrome *		
Gen-Panel: ID088.04, 12 Gene (26,3 kb)		
ELANE, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Primäre Immundefekte, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID380.01, 435 Gen (930,7 kb)		
ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALPI, ALPK1, ANGPT1, ANKZF1, AP1S3, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATAD3A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C20RF69, C3, C5, C6, C7, C8A, C8B, C9, CARD10, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CBLB, CCBE1, CD19, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFHR1, CFI, CFP, CFTR, CHD7, CIB1, CIITA, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNNBL1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DPP9, DUT, EFL1, ELANE, ELF4, EPG5, ERBIN, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FGL2, FLT3LG, FMNL2, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC3, G6PD, GATA1, GATA2, GFI1, GIMAP5, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HPS1, HPS4, HPS6, HSPA1L, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1R1, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IPO8, IRAK4, IRF1, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, IVNS1ABP, JAGN1, JAK1, JAK3, KCNA5, KDM6A, KMT2A, KMT2D, KRAS, LACC1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYN, LYST, MAGT1, MALT1, MAP3K14, MBL2, MCM10, MCM4, MCTS1, MECOM, MEFV, MOGS, MPEG1, MPO, MRTFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NPC1, NRAS, NSMCE3, NUDCD3, OAS1, ORAI1, OTULIN, PARN, PAX1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLG, PNP, POLA1, POLD1, POLD3, POLE, POLR3A, POLR3C, POLR3F, POMP, PRF1, PRIM1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSTPIP1, PTCRA, PTEN, PTPN2, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RC3H1, RECQL4, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHBDF2, RHOH, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCGN, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLC9A3, SLCO2A1, SMARCAL1, SMARCD2, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STX11, STXBP2, STXBP3, SYK, TAP1, TAP2, TAPBP, TAFAZZIN, TBK1, TBX1, TBX21, TCF3, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TGFBR1, TGFBR2, TICAM1, TINF2, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TOM1, TOP2B, TPP2, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC7A, TYK2, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, XIAP, ZAP70, ZBTB24, ZNF341, ZNFX1		
Schwerer kombinierter Immundefekt (SCID) *	3 - 5 Wo	E
Gen-Panel: ID381.00, 21 Gene (47,6 kb)		
ADA, AK2, BCL11B, CD3E, CD3D, CIITA, CORO1A, DCLRE1C, FOXN1, IL2RG, IL7R, LAT, LIG4, JAK3, NHEJ1, POLD3, PRKDC, PTPRC, RAC2, RAG1, RAG2		
Thrombotische Mikroangiopathie (TMA) *	3 - 5 Wo	E
Gen-Panel: ID707.00, 23 Gene (44,7 kb)		
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD		
Lungen- und Respiratorische Erkrankungen		
Chronische Granulomatose (CGD) *	3 - 5 Wo	E
Gen-Panel: ID379.00, 8 Gene (8,8 kb)		
CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2		
Interstitielle Lungenerkrankung und Lungenfibrose *	3 - 5 Wo	E
Gen-Panel: ID341.01		
Interstitielle Lungenerkrankung und Lungenfibrose: 40 Gene (80,9 kb)		
ABCA3*, ACD, AP3B1, AP3D1, COPA, CSF2RA, CSF2RB, DCLRE1B, DKC1, FAM111B, FARSA, FARSB, FLNA, FOXF1, GBA1, HPS1, HPS4, ITGA3, MARS1, NAF1, NHP2, NKX2-1, NOP10, OAS1, PARN, RPA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STING1, TERC, TERT, TINF2, WRAP53, ZCCHC8		
Pulmonale Alveolarproteinose (SMDP, ILD): 10 Gene (17,7 kb)		
ABCA3*, CSF2RA, CSF2RB, MARS1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC7A7, OAS1		
Telomer-assoziierte Lungenfibrose (PFBMFT, DKC): 14 Gene (22,7 kb)		
ACD, DCLRE1B, DKC1, NAF1, NHP2, NOP10, PARN, RPA1, RTEL1, TERC, TERT, TINF2, WRAP53, ZCCHC8		
Syndrome mit Lungenfibrose: 16 Gene (32,6 kb)		
ACD, AP3B1, AP3D1, DKC1, FAM111B, HPS1, HPS4, NAF1, NHP2, NOP10, PARN, RTEL1, SLC34A2, TERT, TINF2, WRAP53		
Syndrome mit diffuser parenchymaler Lungenerkrankungen: 10 Gene (25,0 kb)		
COPA, FARSA, FARSB, FLNA, FOXF1, GBA1, ITGA, NKX2-1, SMPD1, STING1		
Kongenitales zentrales Hypoventilationssyndrom (CCHS) *	3 - 5 Wo	E
Gen-Panel: ID185.01, 5 Gene (9,7 kb)		
LBX1, MECP2, MYO1H, PHOX2B, RET		
Lungenkarzinom *	4 - 6 Wo	E
Gen-Panel: ID260.02, 33 Gene (122,5 kb)		
ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTYH, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53		
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *	4 - 6 Wo	E
Gen-Panel: ID085.03, 50 Gene (164,8 kb)		
BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10		
Pulmonale Hypertonie (PAH) *	3 - 5 Wo	E
Gen-Panel: ID281.01, 23 Gene (49,9 kb)		
ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4		
Pulmonale Surfactant-Stoffwechselstörung (SMDP) *	3 - 5 Wo	E
Gen-Panel: ID168.01, 6 Gene (12,0 kb)		
ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB		
Zystische Fibrose (CF) *	3 - 5 Wo	E
Gen-Panel: ID045.00, 1 Gen (4,4 kb)		
CFTR		

Erkrankung/Diagnostik	Dauer	Material
Mikrozephalien und Makrozephalien		
Galloway-Mowat-Syndrom (GAMOS) *	3 - 5 Wo	E
Gen-Panel: ID251.01, 10 Gene (12,5 kb)		
GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC		
Hydrozephalie *	3 - 5 Wo	E
Gen-Panel: ID221.04		
Hydrozephalie: 20 Gene (57,9 kb)		
AKT3, CCDC88C, CCND2, CFAP43, FANCB, FLVCR2, FOXJ1, GPSM2, HYLS1, KIF7, L1CAM, MPDZ, P4HB, PIK3R2, POMT1, SEC24D, SMARCC1, TRIM71, WDR81, ZIC3		
Kongenitaler Hydrozephalus (HYC): 6 Gene (27,7 kb)		
CCDC88C, L1CAM, MPDZ, SMARCC1, TRIM71, WDR81		
Syndrome mit Hydrozephalus: 14 Gene (29,0 kb)		
AKT3, CCND2, FANCB, FLVCR2, FOXJ1, GPSM2, HYLS1, KIF7, L1CAM, P4HB, PIK3R2, POMT1, SEC24D, ZIC3		
Intellektuelle Entwicklungsstörung und Makrozephalie *	4 - 6 Wo	E
Gen-Panel: ID131.03		
Intellektuelle Entwicklungsstörung und Makrozephalie: 48 Gene (147,3 kb)		
ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Makrozephalie: 21 Gene (70,8 kb)		
CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Makrozephalie: 10 Gene (20,2 kb)		
ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMCO1, ZBTB7A		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Makrozephalie: 18 Gene (57,9 kb)		
BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9		
Intellektuelle Entwicklungsstörung und Mikrozephalie *	4 - 6 Wo	E
Gen-Panel: ID129.02		
Intellektuelle Entwicklungsstörung und Mikrozephalie: 80 Gene (229,8 kb)		
ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNNB1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNPH2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Mikrozephalie: 32 Gene (100,5 kb)		
AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Mikrozephalie: 21 Gene (41,0 kb)		
ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Mikrozephalie: 27 Gene (88,3 kb)		
ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2		
Mikrozephalie, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID069.02		
Mikrozephalie, umfassende Diagnostik: 129 Gene (371,7 kb)		
ADARB1, ANKLE2, ARCN1, ARFGEF2, ASPM, ATR, BLM, CARS1, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CENPT, CEP135, CEP152, CEP63, CIT, CKAP2L, COPB1, COPB2, CRIPT, CTNNB1, CTU2, DIAPH1, DNA2, DNMT3A, DONSON, DPP6, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, ERCC1, ERCC2, ERCC5, ERCC6, EXOC8, EXT2, FOXG1, GEMIN4, GPT2, IER3IP1, KAT6A, KATNB1, KCNA4, KIF11, KIF14, KNL1, LAGE3, LMNB1, LMNB2, MCPH1, MED17, MFSD2A, MSMO1, MTHFS, MYCN, NARS1, NBN, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIN, NSMCE2, NUP107, NUP133, NUP37, OCLN, OSGEP, PCDH12, PCNT, PHC1, PHGDH, PLAA, PLEKHG2, PLK4, PNKP, PPP1R15B, PQBP1, PRUNE1, PSAT1, PUS3, PUS7, QARS1, RAB18, RAB3GAP1, RAB3GAP2, RAD50, RBBP8, RTTN, SARS1, SASS6, SLC1A4, SLC25A19, SLC9A6, SMPD4, SPOP, STAG2, STAMBP, STIL, SVBP, TBC1D20, THOC6, TMX2, TOP3A, TP53RK, TPRKB, TRAIP, TRAPPC6B, TRAPPC14, TRIO, TRMT10A, TUBGCP2, TUBGCP4, TUBGCP6, VARS1, VPS13B, WDFY3, WDR4, WDR62, WDR73, XRCC4, YIPF5, ZEB2, ZNF335		
Primäre Mikrozephalie (MCPH): 27 Gene (110,8 kb)		
ANKLE2, ASPM, CDK6, CIT, CDK5RAP2, CENPE, CENPJ, CEP135, CEP152, COPB2, KIF14, KNL1, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PHC1, SASS6, STIL, TRAPPC14, WDFY3, WDR62, ZNF335		
Seckel-Syndrom (SCKL): 9 Gene (33,5 kb)		
ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP		
Warburg-Mikro-Syndrom (WARBM): 4 Gene (8,9 kb)		
RAB18, RAB3GAP1, RAB3GAP2, TBC1D20		
Galloway-Mowat-Syndrom (GAMOS): 8 Gene (11,4 kb)		
LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73		
Zerebrookulofazioskelettales Syndrom (COFS): 4 Gene (11,3 kb)		
ERCC1, ERCC2, ERCC5, ERCC6		
Mikrozephalie und Chorioretinopathie (MCCRP): 4 Gene (13,5 kb)		
PLK4, TUBGCP4, TUBGCP6, KIF11		
Neurologische Entwicklungsstörung mit Mikrozephalie (NEDM): 21 Gene (39,7 kb)		
ADARB1, COPB1, CTNNB1, DYNC1I2, EXOC8, GEMIN4, GPT2, MFSD2A, MTHFS, NARS1, PLAA, PRUNE1, PUS3, SPOP, STAG2, TMX2, TRAPPC6B, SARS1, SMPD4, SVBP, VARS1		

Erkrankung/Diagnostik	Dauer	Material
Makrozephalie, umfassende Diagnostik * Gen-Panel: ID070.03 Makrozephalie, umfassende Diagnostik: 135 Gene (364.6 kb) ABCC9, ADK, AKT1, AKT3, ALKBH8, AMER1, ANKH, APC2, ASPA, ASXL2, BRAF, BRWD3, CAMK2G, CCDC22, CCDC88C, CCND2, CDH2, CDKN1C, CHD1, CHD3, CHD8, CRADD, CSF1R, CUL4B, DIS3L2, DDX3X, DEAF1, DNMT3A, DVL1, DVL3, EED, EML1, EXT2, EZH2, FGFR3, FIBP, FMR1, FOXP1, GATAD2B, GCDH, GFAP, GLI3, GPC3, GPC4, GRIA3, GUSB, H1-4, HEPACAM, HERC1, HRAS, HSD17B4, HUWE1, IGBP1, INPPL1, KDM5C, KIF7, KPTN, KRAS, L1CAM, LAMB1, LBR, LZTR1, MAN2B1, MAP2K1, MAP2K2, MAPK1, MECP2, MED12, MITF, MLC1, MPDZ, MRAS, MSL3, MTOR, NF1, NFIB, NFIX, NONO, NRAS, NSD1, NXN, OFD1, OPHN1, PAK1, PHF21A, PIGA, PIGM, PIGN, PIGT, PIK3CA, PIK3R2, PKDCC, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, PTPN11, RAB39B, RAC1, RAF1, RIN2, RIT1, RNF125, RNF135, ROR2, RRAS2, SEC23B, SETD2, SHANK3, SHOC2, SHROOM4, SNX10, SOS1, SOS2, SPOP, SPRED1, SPRED2, STRADA, SUFU, SUZ12, TBC1D7, TCIRG1, TMCO1, TNFRSF11A, TRIO, TRIP12, UPF3B, WASHC5, WDR81, WNT5A, ZBTB7A, ZBTB20, ZBTB42, ZDHHC9 Cowden-Syndrom (CWS): 4 Gene (8,2kb) AKT1, PIK3CA, PTEN, SEC23 Robinow-Syndrom (RRS, DRS): 5 Gene (9,5 kb) DVL1, DVL3, NXN, ROR2, WNT5A Großwuchssyndrome: 15 Gene (38,9 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, FIBP, GPC3, GPC4, NFIX, NSD1, OFD1, PTEN, RNF125, SETD2, SUZ12 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Intellektuelle Entwicklungsstörungen und Makrozephalie: 49 Gene (147,9 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, H1-4, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9	4 - 6 Wo	E
Pontozerebelläre Hypoplasie (PCH) * Gen-Panel: ID071.02, 26 Gene (60,5 kb) AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34 TSEN54, VPS51, VPS53, VRK1	3 - 5 Wo	E
Primäre Mikrozephalie, autosomal-rezessiv (MCPH) * Gen-Panel: ID031.02, 25 Gene (107,1 kb) ANKLE2, ASPM, CDK6, CIT, CDK5RAP2, CENPE, CENPJ, CEP135, CEP152, COPB2, KIF14, KNL1, MAP11, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PHC1, SASS6, STIL, WDFY3, WDR62, ZNF335	4 - 6 Wo	E
Seckel-Syndrom (SCKL) * Gen-Panel: ID113.00, 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP	3 - 5 Wo	E
Wachstumsstörung und Makrozephalie * Gen-Panel: ID072.03 Wachstumsstörung und Makrozephalie: 30 Gene (80,9 kb) AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12 Großwuchssyndrom und Makrozephalie: 11 Gene (33,0 kb) DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12 Noonan-Syndrom und Makrozephalie: 10 Gene (15,2 kb) BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1	3 - 5 Wo	E
Mitochondriopathien		
Coenzym-Q10-Mangel (COQ10D) * Gen-Panel: ID225.01, 15 Gene (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2	3 - 5 Wo	E
Defizienz der mitochondrialen Komplexe I bis V (MCDN) * Panel: ID074.03 Defizienz der mitochondrialen Komplexe I bis V (MCDN): 83 Gene (60,7 kb) ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX5A, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, CYC1, DNAJC30, FOXRED1, LRPPRC, LYRM7*, MTFMT, NDUFA1, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, PET100, PET117, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SURF1, TACO1, TIMMDC1, TMEM70, TMEM126B, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ Mitochondrialer Komplex-I-Mangel, kernkodierter Typ (MC1DN): 39 Gene (29,3 kb) ACAD9, DNAJC30, FOXRED1, MTFMT, NDUFA1, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, TIMMDC1, TMEM126B Mitochondrialer Komplex-II-Mangel, kernkodierter Typ (MC2DN): 4 Gene (3,7 kb) SDHA, SDHAF, SDHB, SDHD Mitochondrialer Komplex-III-Mangel, kernkodierter Typ (MC3DN): 10 Gene (7,1 kb) BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1 Mitochondrialer Komplex-IV-Mangel, kernkodierter Typ (MC4DN): 23 Gene (15,8 kb) COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, COX5A, LRPPRC, NDUFA4, PET100, PET117, SCO1, SCO2, SURF1, TACO1 Mitochondrialer Komplex-V-(ATP-Synthase-)Mangel, kernkodierter Typ (MC5DN): 7 Gene (4,8 kb) ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, TMEM70	3 - 5 Wo	E
Kombinierter oxidativer Phosphorylierungsmangel (COXPD) * Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 Wo	E
Lebersche hereditäre Optikusneuropathie (LHON) [CO] Gen-Panel: ID701.00, 17 Gene (10,5 kb) MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT-TQ, MT-TT	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
MELAS-Syndrom [CO] Gen-Panel: ID700.01, 22 Gene (9,2 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-RNR2, MT-TC, MT-TE, MT-TF, MT-TH, MT-TK, MT-TL1, MT-TL2, MT-TQ, MT-TS1, MT-TS2, MT-TV, MT-TW	3 - 5 Wo	E
Mitochondriales DNA-Depletionssyndrom (MTDPS) * Gen-Panel: ID324.01, 21 Gene (29,3 kb) AGK, DGUOK, FBXL4, GUK1, LIG3, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP	3 - 5 Wo	E
Mitochondriale Myopathie / Enzephalopathie [CO] Gen-Panel: ID702.00, 27 Gene (7,0 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND2, MT-ND5, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TW	3 - 5 Wo	E
Mitochondriengenom [CO] Gen-Panel: ID703.00, 37 Gene (11,4 kb) MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY	3 - 5 Wo	E
Nukleär-kodierte mitochondriale Erkrankungen * Gen-Panel: ID704.01, 312 Gene (406,1 kb) AARS2, ABCB7, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH4A1, ALDH6A1, AMACR, AMT, ANO10, APTX, ATL1, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATPAF2, AUH, BCAT2, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C1QBP, CARS2, CISD2, COA3, COA5, COA6, COA8, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX5A, COX6A2, COX6B1, CPS1, CPT1A, CPT2, CYB5R3, CYC1, CYCS, CYP27A1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, EARS2, ELAC2, ERCC6, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBP1, FBXL4, FH, FOXRED1, FXN, GAMT, GARS1, GATB, GATC, GATM, GCDH, GCK, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GTPBP3, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HK1, HLCS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSPD1, HTRA2, IDH1, IDH2, IDH3B, ISCU, IVD, KARS1, KIF1B, KIF5A, L2HGDH, LARS2, LIAS, LRPPRC, LYRM4, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICOS13, MIEF2, MIPEP, MLYCD, MMAA, MMAB, MMADHC, MMUT, MPV17, MRM2, MRPL12, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS28, MRPS34, MRPS7, MTFMT, MTO1, MTPAP, MTRFR, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NSUN3, NUBPL, OAT, OPA1, OPA3, OTC, OXCT1, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PET117, PINK1, PNKD, PNPLA2, PNPT1, POLG, POLG2, PPOX, PRKN, PTCD3, PUS1, QRSL1, RARS2, REEP1, RMND1, RMRP, RNASEH1, RRM2B, SACS, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A10, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC33A1, SLC6A8, SPART, SPAST, SPG7, STAR, SUCLA2, SUCLG1, SURF1, TACO1, TAFAZZIN, TARS2, TFAM, TIMM22, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UNG, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, VARS2, WFS1, WWOX, XPNPEP3, YARS2, YWHAE	4 - 6 Wo	E
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB) * Gen-Panel: ID300.00 Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB): 10 Gene (17,9 kb) DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK Progressive externe Ophthalmoplegie, autosomal-dominant (PEOA): 6 Gene (12,4 kb) DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK Progressive externe Ophthalmoplegie, autosomal-rezessiv (PEOB): 5 Gene (9,2 kb) DGUOK, POLG, RNASEH1, TK2, TOP3A	3 - 5 Wo	E
Multisystem-Fehlbildungssyndrome		
Alagille-Syndrom (ALGS) * Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
Alport-Syndrom (ATS) * Gen-Panel: ID099.00, 4 Gene (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 Wo	E
Bardet-Biedl-Syndrom (BBS) * Gen-Panel: ID093.02, 21 Gene (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 Wo	E
CHARGE-Syndrom * Gen-Panel: ID307.00, 3 Gene (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 Wo	E
Coffin-Siris-Syndrom (CSS) * Gen-Panel: ID118.02, 14 Gene (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 Wo	E
Cornelia-de-Lange-Syndrom (CDLS) * Gen-Panel: ID033.02, 8 Gene (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 Wo	E
Fehlbildungssyndrome mit überwiegend fazialer Beteiligung * Gen-Panel: ID279.00 Fehlbildungssyndrome mit überwiegend fazialer Beteiligung: 25 Gene (90,9 kb) C2CD3, COL11A2, CPLANE1, DDX59, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GRIP1, IFT57, INTU, KIAA0753, MEGF8, MYH3, MYMK, MYT1, OFD1, RAB23, RBM10, TCTN3, TGDS, TMEM107, TNNI2, TWIST1 Akrozephalosyndaktylie-Syndrom (ACS): 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107 Fraser-Syndrom (FRASRS): 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1 Pierre-Robin-Syndrom: 4 Gene (9,7 kb) COL11A2, MYMK, RBM10, TGDS	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
FG-Syndrom (FGS) *	3 - 5 Wo	E
Gen-Panel: ID215.00, 3 Gene (17,2 kb)		
CASK, FLNA, MED12		
Kabuki-Syndrom (KABUK) *	3 - 5 Wo	E
Gen-Panel: ID127.00, 2 Gene (20,8 kb)		
KDM6A, KMT2D		
Kombinierter oxidativer Phosphorylierungsmangel (COXPD) *	3 - 5 Wo	E
Gen-Panel: ID287.01, 60 Gene (80,3 kb)		
AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2		
Kongenitale Glykosylierungsstörung (CDG) *	3 - 5 Wo	E
Gen-Panel ID035.03		
Kongenitale Störung der Glykosylierung (CDG): 58 Gene (86,2 kb)		
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, GET4, MAGT1, MAN1B1, MAN2B2, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, STX5, TMEM165, TUSC3, VMA12, VMA22		
Kongenitale Glykosylierungsstörung, Typ I (CDG1): 31 Gene (43,6 kb)		
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAN2B2, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3		
Kongenitale Glykosylierungstörung, Typ II (CDG2): 27 Gene (42,7 kb)		
ATP6AP1, ATP6AP2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, GET4, MAN1B1, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, STX5, TMEM165, VMA12, VMA22		
Marfan-Syndrom (MFS) *	2 - 4 Wo	E
Gen-Panel: ID022.00, 3 Gene (11,8 kb)		
FBN1 TGFBR1, TGFBR2		
Noonan-Syndrom (NS) *	3 - 5 Wo	E
Gen-Panel: ID023.06, 16 Gene (27,4 kb)		
BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2		
Orofaziodigitales Syndrom (OFD) *	3 - 5 Wo	E
Gen-Panel: ID265.01, 14 Gene (40,2 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TBC1D32, TCTN3, TMEM107, ZRSR2		
RASopathien *	3 - 5 Wo	E
Gen-Panel: ID015.05		
RASopathien: 21 Gene (40,1 kb)		
BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2		
Noonan-Syndrom (NS): 15 Gene (24,7 kb)		
BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2		
Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb)		
BRAF, KRAS, MAP2K1, MAP2K2		
LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb)		
BRAF, PTPN11, RAF1		
Rubinstein-Taybi-Syndrom (RSTS) *	3 - 5 Wo	E
Gen-Panel: ID142.01, 3 Gene (24,3 kb)		
CREBBP, EP300, SRCAP		
Tuberöse Sklerose (TSC) *	2 - 4 Wo	E
Gen-Panel: ID332.00, 2 Gene (8,9 kb)		
TSC1, TSC2		
VACTERL-Assoziation *	3 - 5 Wo	E
Gen-Panel: ID258.02, 27 Gene (72,8 kb)		
BRCA2, CHD7, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGF8, FOXF1, GLI3, HAAO, HOXD13, HSPA6, MNX1, RECQL4, SALL1, KYNU, MYCN, NADSYN1, RAD51C, TRAP1, WBP11, ZIC3		
Weill-Marchesani-Syndrom (WMS) *	3 - 5 Wo	E
Gen-Panel: ID230.00, 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Zellweger-Syndrom (ZWS) *	3 - 5 Wo	E
Gen-Panel: ID084.00, 14 Gene (22,3 kb)		
ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Alzheimer-Krankheit (AD) *	3 - 5 Wo	E
Gen-Panel: ID157.02, 9 Gene (23,4 kb)		
A2M, ABCA7, ADAM10, APOE, APP, NOS3, PSEN1, PSEN2, TREM2		
Amyotrophe Lateralsklerose (ALS) *	3 - 5 Wo	E
Gen-Panel: ID209.04, 35 Gene (80,2 kb)		
ALS2, ANG, ANXA11, ATXN2, C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, LRP12, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SPTLC1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP		
Basalganglien-Kalzifikation (IBGC) *	3 - 5 Wo	E
Gen-Panel: ID327.00, 6 Gene (11,2 kb)		
JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1		
CADASIL und CARASIL *	3 - 5 Wo	E
Gen-Panel: ID167.01, 3 Gene (9,4 kb)		
HTRA1, NOTCH3, TREX1		
Choreatiforme Bewegungsstörungen *	3 - 5 Wo	E
Gen-Panel: ID272.01, 17 Gene (40,6 kb)		
ADCY5, ATP1A2, ATP1A3, FRRS1L, GNAO1, HPRT1, KCNMA1, NKX2-1, PDE2A, PDE10A, PNKD, PRNP, PRRT2, RNF216, SLC2A1, VPS13A, XK		
Distale motorische Neuronopathie (HMND, HMNR, HMNX) *	3 - 5 Wo	E
Gen-Panel: ID254.02		
Distale motorische Neuronopathie (HMND, HMNR, HMNX): 26 Gene (58,7 kb)		
ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
Distale motorische Neuronopathie, autosomal-dominant (HMND): 16 Gene (40,2 kb)		
BAG3, BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1		
Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 11 Gene (19,2 kb)		
ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, RTN1, SIGMAR1, SORD, VRK1, VWA1		
Dystonie (DYT) *	3 - 5 Wo	E
Gen-Panel ID128.04		
Dystonie (DYT): 41 Gene (84,3 kb)		
ACTB, ADCY5, ANO3, AOPEP, ATP1A3, ATP5MC3, BCAP31, COL6A3, ECHS1, EIF2AK2, GCH, GNAL, HPCA, KCNN2, KCTD17, KMT2B, MECR, NUP54, PLA2G6, PNKD, PRKRA, PRRT2, SGCE, SHQ1, SLC2A1, SLC6A3, SLC18A2, SLC30A10, SLC39A14, SPR, TAF1, TH, THAP1, TIMM8A, TMEM151A, TOR1A, TSPOAP1, TUBB4A, VPS11, VPS16, WARS2		
Dystonie (DYT), isolierte Form: 15 Gene (39,4 kb)		
ADCY5, ANO3, COL6A3, ECHS1, GNAL, HPCA, KCNN2, KCTD17, KMT2B, SHQ1, SLC2A1, THAP1, TOR1A, TUBB4A, VPS11		
Dystonie (DYT), kombinierte Form: 29 Gene (47,2 kb)		
ACTB, ADCY5, AOPEP, ATP1A3, ATP5MC3, BCAP31, ECHS1, EIF2AK2, GCH1, MECR, NUP54, PLA2G6, PNKD, PRKRA, PRRT2, SGCE, SHQ1, SLC2A1, SLC6A3, SLC18A2, SLC30A10, SLC39A14, SPR, TAF1, TH, TIMM8A, TMEM151A, VPS16, WARS2		
Episodische Ataxie (EA) *	3 - 5 Wo	E
Gen-Panel: ID184.02, 6 Gene (20,6 kb)		
ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3		
Essentieller Tremor (ETM) *	3 - 5 Wo	E
Gen-Panel: ID195.01, 4 Gene (16,6 kb)		
DRD3, FUS, SCN4A, TENM4		
Familiäres episodisches Schmerzsyndrom (FEPS) *	3 - 5 Wo	E
Gen-Panel: ID268.01, 6 Gene (22,3 kb)		
GLA, SCN9A, SCN10A, SCN11A, TRPA1, TTR		
Frontotemporale Demenz (FTD) *	3 - 5 Wo	E
Gen-Panel: ID310.02, 19 Gene (29,3 kb)		
C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP		
Hereditäre Ataxien, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID276.03		
Hereditäre Ataxien, umfassende Diagnostik: 128 Gene (373,1 kb)		
ABCB7, ABHD12, ACO2, AFG3L2, ANGPTL1, AIFM1, ANO10, APOB, APTX, ATCAY, ATG5, ATG7, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CASK, CCDC88C, CHP1, CLCN2, CLN5, COA7, COQ2, COQ4, COQ8A, CWF19L1, CYP27A1, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, FLVCR1, GDAP2, GOSR2, GRID2, GRM1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, LAMA1, MARS2, MME, MRE11, MTCL1, MTPAP, MTTP, NBN, NKX6-2, NPTX1, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PITRM1, PLD3, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRDX3, PRKCG, PRPS1, PTF1A, PUM1, RFC1, RNF216, RNU12, RUBCN, SACS, SAMD9L, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC2A1, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VPS41, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1		
Episodische Ataxie (EA): 6 Gene (20,5 kb)		
ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3		
Spastische Ataxie (SPAX): 7 Gene (25,6 kb)		
AFG3L2, CHP1, COQ4, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1		
Spinozerebelläre Ataxie, autosomal-dominant (SCA): 27 Gene (91,6 kb)		
AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPTX1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2		
Spinozerebelläre Ataxie, autosomal-rezessiv (SCAR, SCAN): 32 Gene (106,7 kb)		
ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, UBA5, VPS13D, VPS41, VWA3B, WWOX, XRCC1		
Zerebelläre Ataxie mit mentaler Retardierung (CAMRQ): 4 Gene (12,9 kb)		
ATP8A2, CA8, VLDLR, WDR81		
Zerebelläre Ataxie, X-chromosomal: 7 Gene (15,9 kb)		
ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6		
Ataxie mit okulomotorische Apraxie (AOA): 4 Gene (13,3 kb)		
APTX, PIK3R5, PNKP, SETX		
Ataxia teleangiectatica (AT): 5 Gene (15,4 kb)		
APTX, ATM, MRE11, NBN, PCNA		

Erkrankung/Diagnostik	Dauer	Material
Hereditäre Neuropathien (HMSN, HMN, HSAN), umfassende Diagnostik * Gen-Panel: ID374.01 Hereditäre Neuropathien (HMSN, HMN, HSAN), umfassende Diagnostik: 136 Gene (347,7 kb) AARS1, ABCA1, ACOX1, AIFM1, AR, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CADM3, CFAP276, CHCHD10, CNTNAP1, COQ7, COX20, COX6A1, CPOX, CYP27A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, GSN, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MAG, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PDXK, PIGB, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PPOX, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, RTN1, SBF1, SBF2, SCN9A, SCN10A, SCN11A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A6, SLC5A7, SLC52A2, SLC52A3, SMN1, SORD, SPG11, SPTAN1, SPTLC1, SPTLC2, SURF1, SYT2, TECPR2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VCP, VRK1, VWA1, WARS1, WNK1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2, HMSN2): 35 Gene (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX, HMSN): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophe Déjerine-Sottas-Neuropathie (DSS, HMSN3): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb) EGR2, MPZ, CNTNAP1 Distale motorische Neuropathie (HMND, HMNR, HMNX): 26 Gene (58,7 kb) ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Sensorische und autonome Neuropathie (HSAN, HSN): 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	4 - 6 Wo	E
Hereditäre sensorische und autonome Neuropathie (HSAN, HSN) * Gen-Panel: ID086.02, 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	3 - 5 Wo	E
Hirnatrophie und demyelinisierende Erkrankungen des Gehirns * Gen-Panel: ID278.00 Hirnatrophie und demyelinisierende Erkrankungen des Gehirns: 55 Gene (96,0 kb) AIMP1, AIMP2, ASPA, B3GALNT2, B4GAT1, CNP, COL4A1, CRPPA, DAG1, DARS1, DEGS1, EPRS1, FAM126A, FARSA, FARSB, FKRP, FKTN, GFAP, GJC2, GMPPB, GRM7, HIKESHI, HSPD1, LARGE1, MAPT, MAT1A, MED17, MTHFS, PLP1, POLR1C, POLR3A, , POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSEN1, PYCR2, RARS1, RXYLT1, SLC25A12, SOX10, TBCD, TMEM106B, TMEM63A, TRAPPC12, TRAPPC4, TRAPPC6B, UBTF, UFM1, VPS11 Hirnatrophie: 13 Gene (24,2 kb) EXOC7, EXOC8, FARSA, FARSB, GRM7, MAPT, MED17, PSEN1, TBCD, TRAPPC4, TRAPPC6B, TRAPPC12, UBTF Hypo- und Demyelinisierung des Gehirns: 27 Gene (43,0 kb) AIMP1, AIMP2, ASPA, CNP, DARS1, DEGS1, EPRS1, FAM126A, GFAP, GJC2, HIKESHI, HSPD1, MAT1A, MTHFS, PLP1, POLR1C, POLR3A, POLR3B, PYCR2, RARS1, SLC25A12, SOX10, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Walker-Warburg-Syndrom (MDDGA): 14 Gene (23,6 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Hyperekplexie (HKPX) * Gen-Panel: ID216.00, 9 Gene (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 Wo	E
Hypomyelinisierende Leukodystrophie (HLD) * Gen-Panel: ID277.00, 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11	3 - 5 Wo	E
Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik * Gen-Panel: ID204.04 Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik: 122 Gene (208,5 kb) AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, AIFM1, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, AUH, BOLA3, CLCN2, CLDN11, CNP, COA7, COA8, COL4A1, COL4A2, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS1, DARS2, DEGS1, EARS2, EIF2AK1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FA2H, FAM126A, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GCDH, GFAP, GFM1, GJC2, GLB1, HEPACAM, HIKESHI, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, ISCA1, ISCA2, KARS1, KCNT1, L2HGDH, LMNB1, LYRM7, MARS2, MCOLN1, MLC1, MTFMT, NAXD, NAXE, NFU1, NKX6-2, NOTCH3, NUBPL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PLAA, PLEKHG2, PLP1, PMPCB, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SDHAF1, SLC13A3, SLC16A2, SLC17A5, SOX10, SPTAN1, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYMP, TYROBP, UFM1, VPS11, ZFYVE26 Leukodystrophie mit Hypomyelinisierung (HLD): 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Leukodystrophie mit Peroxisomenbiogenese-Störung (PBD): 15 Gene (20,8 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B*, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH Orthochromatische Leukodystrophie: 10 Gene (14,6 kb) ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1 Metachromatische Leukodystrophie: 3 Gene (4,2 kb) ARSA, PSAP, SUMF1 Aicardi-Goutières-Syndrom (AGS): 7 Gene (11,9 kb) ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1 CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Komplexe neurologische Entwicklungsstörungen (NED) *	4 - 6 Wo	E
Gen-Panel: ID358.01, 193 Gene (496,0 kb)		
ACBD6, ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BAZ2B, BCAS3, BPTF, BRAT1, C18ORF32, CAPRIN1, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CERT1, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DPH5, DYNC1I2, EEF1D, EMC10, ESAM, EXOC2, EXOC7, EXOC8, FAM177A1, FBXW11, FDFT1, FEM1B, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, GTPBP2, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HDAC4, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCND1, KCNN2, KDM5A, KDM6B, LNPK, MADD, MAP4K4, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NAV3, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPMT1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, RNU4-2, SARS1, SEC31A, SEL1L, SETD1A, SHMT2, SHQ1, SLC4A10, SMG8, SMG9, SMPD4, SNF8, SNIP1, SPOP, SPOUT1, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, TTI1, UBE3C, UBE4A, UBR5, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZBTB11, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6		
Neurodegeneration mit Eisenablagerung im Gehirn (NBIA) *	3 - 5 Wo	E
Gen-Panel: ID264.00, 11 Gene (20,0 kb)		
ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45		
Neuronale Ceroid-Lipofuszinose (CLN) *	3 - 5 Wo	E
Gen-Panel: ID132.01, 15 Gene (20,2 kb)		
ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1		
Parkinson-Krankheit (PARK) und Parkinsonismus*	3 - 5 Wo	E
Gen-Panel: ID077.03		
Parkinson-Krankheit (PARK) und Parkinsonismus: 49 Gene (107,1 kb)		
ATP13A2, ATP1A3, ATP6AP2, C19ORF12, CHCHD2, COASY, DCTN1, DNAJC6, DNAJC12, EIF4G1, FBXO7, FTL, GBA1, GCH1, GIGYF2, GRN, HTRA2, LRRK2, MAPT, NR4A2, PARK7, PANK2, PGK1, PINK1, PLA2G6, POLG, PRKRA, PRKN, PSAP, PTPA, PTRHD1, RAB32, RAB39B, SLC18A2, SLC30A10, SLC39A14, SLC6A3, SLC9A6, SNCA, SYNJ1, TAF1, TH, TRPM7, UCHL1, UQCRC1, VPS13C, VPS35, WARS2, WDR45		
Parkinson-Krankheit (PARK), autosomal-rezessiv: 10 Gene (31,3 kb)		
ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, PTPA, SYNJ1, VPS13C		
Parkinson-Krankheit (PARK), autosomal-dominant: 11 Gene (25,5 kb)		
CHCHD2, EIF4G1, GBA1, GIGYF2, HTRA2, LRRK2, PSAP, RAB32, SNCA, UCHL1, VPS35		
Dystonie-Parkinsonismus (DYT): 12 Gene (23,6 kb)		
ATP1A3, GCH1, NR4A2, PLA2G6, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH, WARS2		
Komplexe Parkinson-Syndrome: 20 Gene (33,9 kb)		
ATP6AP2, C19ORF12, COASY, DCTN1, DNAJC12, FTL, GRN, MAPT, NR4A2, PANK2, PGK1, POLG, PTRHD1, RAB39B, SLC9A6, SLC30A10, SLC39A14, TRPM7, UQCRC1, WDR45		
Paroxysmale Dyskinesie *	3 - 5 Wo	E
Gen-Panel: ID286.00, 14 Gene (38,9 kb)		
ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24		
Schlafstörung *	3 - 5 Wo	E
Gen-Panel: ID371.00, 13 Gene (25,3 kb)		
ADRB1, BHLHE41, CRY1, CRY2, CSNK1D, GRM1, HCRT, MOG, NPSR1, PER2, PER3, PRNP, TIMELESS		
Spastische Ataxie (SPAX) *	3 - 5 Wo	E
Gen-Panel: ID228.00, 12 Gene (34,8 kb)		
AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1		
Spastische Paraplegie (HSP, SPG) *	4 - 6 Wo	E
Gen-Panel: ID148.05		
Spastische Paraplegie (HSP, SPG): 68 Gene (140,5 kb)		
ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, TMEM63C, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-dominant: 20 Gene (43,1 kb)		
ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-rezessiv: 51 Gene (103,6 kb)		
ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, TMEM63C, UCHL1, VPS37A, ZFYVE26		
Spastische Paraplegie (SPG), X-chromosomal: 3 Gene (6,9 kb)		
ABCD1, L1CAM, PLP1		
Spinale Muskelatrophie (SMA) *	3 - 5 Wo	E
Gen-Panel: ID152.03		
Spinale Muskelatrophie (SMA): 41 Gene (91,7 kb)		
AR, ASAH1, ASCC1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1		
Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb)		
AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB		
Distale spinale Muskelatrophie (DSMA, HMN): 26 Gene (58,1 kb)		
ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
Spinozerebelläre Ataxie, autosomal-dominant (SCA, ADCA) *	3 - 5 Wo	E
Gen-Panel: ID236.03, 27 Gene (91,6 kb)		
AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPTX1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2		
Spinozerebelläre Ataxie, autosomal-rezessiv (SCAR, SCAN) *	4 - 6 Wo	E
Gen-Panel: ID213.04, 36 Gene (117,6 kb)		
ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PNPLA6, POLG, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TWNK, UBA5, VPS13D, VPS41, VWA3B, WDR73, WWOX, XRCC1		

Erkrankung/Diagnostik	Dauer	Material
Störung der Peroxisomenbiogenese (PBD) *		
Gen-Panel: ID083.01		
Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	3 - 5 Wo	E
Neonatale Adrenoleukodystrophie / Infantiles Refsum-Syndrom (PBD, Typ B): 11 Gene (17,0 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26		
Heimler-Syndrom (PBD, Typ C): 2 Gene (6,8 kb)		
PEX1, PEX6		
Neuromuskuläre Erkrankungen		
Amyotrophe Lateralsklerose (ALS) *		
Gen-Panel: ID209.04, 35 Gene (80,2 kb)	3 - 5 Wo	E
ALS2, ANG, ANXA11, ATXN2, C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, LRP12, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SPTLC1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP		
Arthrogrypose *		
Gen-Panel: ID200.01		
Arthrogrypose: 61 Gene (209,7 kb)		
ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNM2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2		
Arthrogryposis multiplex congenita (AMC): 6 Gene (58,2 kb)	4 - 6 Wo	E
ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1		
Distale Arthrogrypose (DA): 11 Gene (40,3 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Letales kongenitales Kontraktursyndrom (LCCS): 11 Gene (31,5 kb)		
ADCY6, ADGRG6, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42		
Fetale Akinesie-Deformation-Sequenz (FADS): 4 Gene (7,6 kb)		
DOK7, MUSK, NUP88, RAPSN		
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik *		
Gen-Panel: ID312.02		
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik: 87 Gene (217,3 kb)		
AARS1, AIFM1, ARHGEF10, ATP1A1, ATP7A, BSCL2, CADM3, CNTNAP1, COQ7, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SORD, SPG11, SPTAN1, SPTLC1, SURF1, SYT2, TRIM2, TRPV4, VCP, VRK1, VWA1, WARS1, YARS1		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb)		
EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1		
Charcot-Marie-Tooth-Neuropathie, axonal (CMT2): 35 Gene (97,3 kb)		
AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP		
Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb)	4 - 6 Wo	E
COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1		
Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb)		
AIFM1, GJB1, PDK3, PRPS1		
Hypertrophe Déjerine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb)		
EGR2, MPZ, PMP22, PRX		
Periphere Neuropathie mit Optikusatrophie (HMSN6): 3 Gene (4,5 kb)		
MFN2, PDXK, SLC25A46		
Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb)		
EGR2, MPZ, CNTNAP1		
Distale motorische Neuropathie (HMND, HMNR): 24 Gene (54,7 kb)		
ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN) *		
Gen-Panel: ID052.03		
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN): 45 Gene (111,8 kb)		
AARS1, AIFM1, ATP1A1, CADM3, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, JAG1, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PNKP, PRPS1, RAB7A, SLC12A6, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1		
Charcot-Marie-Tooth-Neuropathie, axonal, dominant (CMT2): 26 Gene (74,2 kb)		
AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, JAG1, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, SLC12A6, TRPV4, VCP	4 - 6 Wo	E
Charcot-Marie-Tooth-Neuropathie, axonal, rezessiv (CMT2): 10 Gene (24,4 kb)		
GDAP1, IGHMBP2, LMNA, LRSAM1, MFN2, MME, MPV17, PNKP, SPG11, TRIM2		
Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb)		
COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1		
Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb)		
AIFM1, GJB1, PDK3, PRPS1		
Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6, HMSN6): 3 Gene (4,5 kb)		
MFN2, PDXK, SLC25A46		

Erkrankung/Diagnostik	Dauer	Material
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN) *	4 - 6 Wo	E
Gen-Panel: ID051.03		
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN): 29 Gene (69,5 kb)		
CNTNAP1, COX6A1, DNM2, EGR2, FBLN5, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, ITPR3, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend, dominant (CMT1): 11 Gene (19,0 kb)		
EGR2, FBLN5, GDAP1, ITPR3, LITAF, MPZ, NEFL, POLR3B, PMP2, PMP22		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-rezessiv (CMT4): 12 Gene (33,8 kb)		
EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1		
Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb)		
COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1		
Hypertrophe Dejerine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb)		
EGR2, MPZ, PMP22, PRX		
Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb)		
EGR, MPZ, CNTNAP1		
Distale Arthrogrypose (DA) *	3 - 5 Wo	E
Gen-Panel: ID196.02, 11 Gene (40,5 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Distale motorische Neuronopathie (HMND, HMNR, HMNX) *	3 - 5 Wo	E
Gen-Panel: ID254.02		
Distale motorische Neuronopathie (HMND, HMNR, HMNX): 26 Gene (58,7 kb)		
ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
Distale motorische Neuronopathie, autosomal-dominant (HMND): 16 Gene (40,2 kb)		
BAG3, BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1		
Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 11 Gene (19,2 kb)		
ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, RTN1, SIGMAR1, SORD, VRK1, VWA1		
Distale Myopathie (MPD) *	4 - 6 Wo	E
Gen-Panel: ID328.01, 30 Gene (186,0 kb)		
ACTA1, ACTN2, ADSS1, ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, HNRNPA1, HNRPA2B1, HSPB8, KY, LDB3, MATR3, MYH7, MYOT, NEB, PYROXD1, SMPX, SQSTM1, TIA1, TCAP, TTN, VCP		
Duchenne- oder Becker-Muskeldystrophie (DMD, BMD) *	3 - 5 Wo	E
Gen-Panel: ID256.00, 1 Gen (11,1 kb)		
DMD		
Emery-Dreifuss-Muskeldystrophie (EDMD) *	3 - 5 Wo	E
Gen-Panel: ID121.00, 6 Gene (51,8 kb)		
EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43		
Gliedergürtelmuskeldystrophie (LGMD) *	4 - 6 Wo	E
Gen-Panel: ID122.04		
Gliedergürtelmuskeldystrophie (LGMD): 37 Gene (196,8 kb)		
ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DNAJB6, DPM3, DYSF, FKRP, FKTN, GMPPB, HNRNPDL, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN		
Gliedergürtelmuskeldystrophie, autosomal-rezessiv (LGMDR): 34 Gene (191,8 kb)		
ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DPM3, DYSF, FKRP, FKTN, GMPPB, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TRAPPC11, TRIM32, TTN		
Gliedergürtelmuskeldystrophie, autosomal-dominant (LGMDD): 7 Gene (23,2 kb)		
CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3		
Gliedergürtelmuskeldystrophie-Dystroglykanopathie (MDDGC): 11 Gene (17,6 kb)		
CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2		
Ullrich-Muskeldystrophie (UCMD): 3 Gene (15,7 kb)		
COL6A1, COL6A2, COL6A3		
Bethlem-Myopathie (BTHLM): 3 Gene (15,7 kb)		
COL6A1, COL6A2, COL6A3		
Kongenitales myasthenes Syndrom (CMS) *	3 - 5 Wo	E
Gen-Panel: ID130.00		
Kongenitales myasthenes Syndrom (CMS): 25 Gene (55,9 kb)		
AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, MYO9A, PREPL, RAPSN, SCN4A, SLC5A7, SLC18A3, SLC25A1, SNAP25, SYT2, VAMP1		
Kongenitales myasthenes Syndrom (CMS), präsynaptisch: 8 Gene (21,9 kb)		
AGRN, CHAT, MYO9A, SLC5A7, SLC18A3, SLC25A1, SYT2, VAMP1		
Kongenitales myasthenes Syndrom (CMS), synaptisch oder postsynaptisch: 18 Gene (40,1 kb)		
AGRN, ALG2, ALG14, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, PREPL, RAPSN, SCN4A, SNAP25		
Kongenitale Myopathie (CMYP) *	4 - 6 Wo	E
Gen-Panel: ID212.02		
Kongenitale Myopathie (CMYP): 50 Gene (269,7 kb)		
ACTA1, ACTN2, BAG3, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, CRYAB, DES, DNAJB4, DNM2, FLNC, FXR1, HACD1, KBTBD13, KLHL40, KLHL41, KY, LDB3, LMOD2, MAP3K20, MEGF10, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYL2, MYOD1, MYOT, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SVIL, TPM2, SELENON, SPEG, STAC3, TNNC2, TNNT1, TPM2, TPM3, TTN, UNC45B		
Nemalin-Myopathie (NEM): 11 Gene (40,6 kb)		
ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3		
Zentronukleäre Myopathie (CNM): 7 Gene (20,3 kb)		
BIN1, CCDC78, DNM2, MAP3K20, MTM1, MTMR14, SPEG		
Myofibrilläre Myopathie (MFM): 12 Gene (130,7 kb)		
BAG3, CRYAB, DES, FLNC, KY, LDB3, MYOT, PYROXD1, TTN		
Central-Core-Myopathie (CCD): 1 Gen (15,1 kb)		
RYR1		

Erkrankung/Diagnostik	Dauer	Material
Hereditäre Neuropathien (HMSN, HMN, HSAN), umfassende Diagnostik * Gen-Panel: ID374.01 Hereditäre Neuropathien (HMSN, HMN, HSAN), umfassende Diagnostik: 136 Gene (347,7 kb) AARS1, ABCA1, ACOX1, AIFM1, AR, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CADM3, CFAP276, CHCHD10, CNTNAP1, COQ7, COX20, COX6A1, CPOX, CYP27A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, GSN, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MAG, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PDXK, PIGB, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PPOX, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, RTN1, SBF1, SBF2, SCN9A, SCN10A, SCN11A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A6, SLC5A7, SLC52A2, SLC52A3, SMN1, SORD, SPG11, SPTAN1, SPTLC1, SPTLC2, SURF1, SYT2, TECPR2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VCP, VRK1, VWA1, WARS1, WNK1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2, HMSN2): 35 Gene (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX, HMSN): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophe Déjerine-Sottas-Neuropathie (DSS, HMSN3): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb) EGR2, MPZ, CNTNAP1 Distale motorische Neuropathie (HMND, HMNR, HMNX): 26 Gene (58,7 kb) ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Sensorische und autonome Neuropathie (HSAN, HSN): 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	4 - 6 Wo	E
Letales kongenitales Kontraktursyndrom (LCCS) * Gen-Panel: ID197.00, 12 Gene (34,6 kb) ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42	3 - 5 Wo	E
Metabolische Muskelerkrankungen und Rhabdomyolyse * Gen-Panel: ID395.00, 93 Gene (205,6 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, AGK, AGL, ALDOA, AMPD1, ANO5, ATP2A2, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHKB, CNTN1, COQ4, COQ8A, CPT2, CTBP1, DMD, DTNA, DNAJB6, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FKRP, FLAD1, GAA, GBE1, GMPPB, GUK1, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LIG3, LPIN1, MGME1, MLIP, MPV17, MRM2, MYH1, OBSCN, OPA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PNPLA2, POC5, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC22A5, SLC25A10, SLC25A20, SLC25A21, SLC25A4, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TK2, TAMM41, TANGO2, TRAPPC2L, TSFM, TWNK, TYMP, YARS2	4 - 6 Wo	E
Muskeldystrophie-Dystroglykanopathie (MDDG) * Gen-Panel: ID179.00 Muskeldystrophie-Dystroglykanopathie (MDDG): 15 Gene (24,0 kb) B3GALNT2, B4GAT1, DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglykanopathie (MDDGA) mit Hirn- und Augenanomalien: 14 Gene (22,8 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglykanopathie (MDDGB) mit oder ohne intellektuelle Entwicklungsstörung: 8 Gene (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2 Muskeldystrophie-Dystroglykanopathie (MDDGC), Gliedergürtelmuskeldystrophie: 11 Gene (18,5 kb) DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, POMGNT1, POMGNT2, POMK, POMT1, POMT2	3 - 5 Wo	E
Muskelerkrankungen, umfassende Diagnostik * Gen-Panel: ID336.00 Muskelerkrankungen, umfassende Diagnostik: 246 Gene (749,8 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, AGRN, ALDOA, ALG14, ALG2, AMPD1, ANO5, ASAH1, ASCC1, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNA2, DNAJB6, DNM2, DNMT3B, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFPT1, GMPPB, GNE, GYG1, GYS1, HADH, HADHA, HADHB, HINT1, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB8, IGHMBP2, INPP5K, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIG3, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MAP3K20, MATR3, MEGF10, MFN2, MGME1, MICU1, MPV17, MPZ, MTM1, MTMR14, MTRFR, MUSK, MYBPC1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, NEFL, OPA1, ORAI1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SCO2, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC52A3, SLC5A7, SMCHD1, SMN1, SNAP25, SPEG, SPG7, SPG11, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TCAP, TFAM, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TRMT5, TRPV4, TSFM, TTN, TUBB3, TWNK, TYMP, UBA1, UNC45B, VAMP1, VCP, VMA21, VRK1, YARS2 Kongenitale, distale und metabolische Myopathien: 110 Gene (382,6 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, ALDOA, AMPD1, ANO5, BAG3, BIN1, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, ECEL1, ENO3, ETFA, ETFB, ETFDH, FLAD1, FLNC, FXR1, GAA, GBE1, GNE, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LDB3, LAMP2, LDHA, LIG3, LMOD3, LPIN1, MAP3K20, MATR3, MEGF10, MGME1, MTM1, MTMR14, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYMK, MYOT, MYPN, NEB, TNNI2, OPA1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, PYROXD1, RBCK1, RRM2B, RYR1, SELENON, SLC22A5, SLC25A20, SLC25A4, SPEG, STAC3, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TIA1, TK2, TNNT1, TNNT3, TPM2, TPM3, TTN, TYMP, UBA1, YARS2 Gliedergürtelmuskeldystrophie (LGMD) und weitere Muskeldystrophien: 47 Gene (270,0 kb) ANO5, B3GALNT2, B4GAT1, BVES, CAPN3, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, HNRNPDL, LAMA2, LARGE1, LIMS2, LMNA, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RXYLT1, SGCA, SGCB, SGC, SGCG, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen mit Herzbeteiligung * Gen-Panel: ID123.03 Muskelerkrankungen mit Herzbeteiligung: 19 Gene (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss-Muskeldystrophie (EMDM): 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker-Muskeldystrophie (DMD, BMD): 1 Gen (11,1 kb) DMD	4 - 6 Wo	E
Myofibrilläre Myopathie (MFM) * Gen-Panel: ID161.01, 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B	4 - 6 Wo	E
Myotonie * Gen-Panel: ID255.00, 5 Gene (25,0 kb) ATP2A1, CLCN1, HINT1, HSPG2, SCN4A	3 - 5 Wo	E
Nemalin-Myopathie (NEM) * Gen-Panel: ID199.00, 11 Gene (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3	3 - 5 Wo	E
Periodische Paralyse * Gen-Panel: ID253.00, 7 Gene (16,6 kb) CACNA1S, KCNE3, KCNJ2, KCNJ5, KCNJ12, KCNJ18, SCN4A	3 - 5 Wo	E
Spastische Paraplegie (HSP, SPG) * Gen-Panel: ID148.05 Spastische Paraplegie (HSP, SPG): 68 Gene (140,5 kb) ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, TMEM63C, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27 Spastische Paraplegie (SPG), autosomal-dominant: 20 Gene (43,1 kb) ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27 Spastische Paraplegie (SPG), autosomal-rezessiv: 51 Gene (103,6 kb) ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, TMEM63C, UCHL1, VPS37A, ZFYVE26 Spastische Paraplegie (SPG), X-chromosomal: 3 Gene (6,9 kb) ABCD1, L1CAM, PLP1	4 - 6 Wo	E
Spinale Muskelatrophie (SMA) * Gen-Panel: ID152.03 Spinale Muskelatrophie (SMA): 41 Gene (91,7 kb) AR, ASAH1, ASCC1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1 Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB Distale spinale Muskelatrophie (DSMA, HMN): 26 Gene (58,1 kb) ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	3 - 5 Wo	E
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Zentronukleäre Myopathie (CNM) * Gen-Panel: ID257.00, 7 Gene (21,4 kb) CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Alport-Syndrom (ATS) * Gen-Panel: ID099.00, 4 Gene (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 Wo	E
Aminoazidurie * Gen-Panel: ID318.00 Aminoazidurie: 13 Gene (21,1 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2 Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Renotubuläres Fanconi-Syndrom (FRTS): 5 Gene (7,7 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC34A1	3 - 5 Wo	E
Atypisches hämolytisch-urämisches Syndrom (AHUS) * Gen-Panel: ID163.04, 20 Gene (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 Wo	E
Bardet-Biedl-Syndrom (BBS) * Gen-Panel: ID093.02, 21 Gene (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 Wo	E
Bartter-Syndrom (BARTS) * Gen-Panel: ID156.01, 8 Gene (17,7 kb) BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3	3 - 5 Wo	E
Branchiootorenales Syndrom (BOR) * Gen-Panel: ID315.00, 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 Wo	E
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen * Gen-Panel: ID706.01, 16 Gene (30,1 kb) ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1	3 - 5 Wo	E
Diabetes insipidus * Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E
Endokrine Hypertonie * Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E
Fraser-Syndrom (FRASRS) * Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
Galloway-Mowat-Syndrom (GAMOS) * Gen-Panel: ID251.01, 10 Gene (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 Wo	E
Glomerulonephritis * Gen-Panel: ID103.01 Glomerulonephritis: 18 Gene (36,9 kb) C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD C3-Glomerulopathie (C3G): 3 Gene (7,2 kb) CFI, CFH, CFHR5 C1q-Mangel (C1QD): 3 Gene (2,3 kb) C1QA, C1QB, C1QC	3 - 5 Wo	E
Hyperoxalurie * Gen-Panel: ID363.00 Hyperoxalurie: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb) AGXT, GRHPR, HOGA1 Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 Wo	E
Hypomagnesiämie (HOMG) * Gen-Panel ID054.02: 14 Gene (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hypophosphatasie, Hypophosphatämie und Rachitis *	3 - 5 Wo	E
Gen-Panel: ID269.03		
Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb)		
ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR		
Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb)		
CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3		
Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb)		
CYP2R1, CYP3A4, CYP27B1, VDR		
Hypophosphatasie (HPP): 1 Gen (1,6 kb)		
ALPL		
Joubert-Syndrom (JBTS) *	4 - 6 Wo	E
Gen-Panel: ID028.03, 40 Gene (104,1 kb)		
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGAGRAM1, TTC21B, ZNF423		
Kongenitale Anomalien der Niere und ableitenden Harnwege (CAKUT) *	4 - 6 Wo	E
Gen-Panel: ID229.03		
Kongenitale Anomalien der Niere und ableitenden Hernwege (CAKUT): 62 Gene (198,3 kb)		
ACE, ACTG2, AGT, AGTR1, ANOS1, BICC1, BMP4, BNC2, CDC5L, CEP55, CHD1L, CHRM3, CRKL, DSTYK, EYA1, FAT4, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3, GFRA1, GREB1L, GRIP1, HNF1B, HPSE2, ITGA8, KIF14, LIFR, LMOD1, LRIG2, LRP4, MUC1, MYH11, MYL9, MYLK, NEK8, NPHP3, NRIP1, PAX2, PBX1, REN, RET, ROBO1, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX11, SOX17, TBC1D1, TBX18, TFAP2A, TNXB, TRAP1, UMOD, UPK3A, WBP11, WNT4		
Renale Hypodysplasie/Aplasie und Agenesie: 25 Gene (75,9 kb)		
ANOS1, BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GFRA1, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4		
Vesikoureteraler Reflux (VUR): 10 Gene (33,7 kb)		
DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB		
Branchiootorenales Syndrom (BOR): 5 Gene (10,2 kb)		
EYA1, SALL1, SIX1, SIX5, TFAP2A		
Renale tubuläre Dysgenesie (RTD): 4 Gene (7,7 kb)		
ACE, AGT, AGTR1, REN		
Fraser-Syndrom (FRARS): 3 Gene (24,8 kb)		
FRAS1, FREM2, GRIP1		
MMIH-Syndrom (MMIHS): 5 Gene (15,1 kb)		
ACTG2, LMOD1, MYH11, MYL9, MYLK		
Meckel-Syndrom (MKS) *	3 - 5 Wo	E
Gen-Panel: ID032.02, 13 Gene (35,1 kb)		
B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231		
Mikrohämaturie *	3 - 5 Wo	E
Gen-Panel: ID385.00, 11 Gene (44,0 kb)		
CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FN1, INF2, MYH9, PIGA, PIGT, UMOD		
Nierensteine und Nephrokalzinose *	3 - 5 Wo	E
Gen-Panel ID231.05		
Nierensteine und Nephrokalzinose: 44 Gene (80,2 kb)		
ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, FAM20A, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, NHERF1, OCRL, OXGR1, PHEX, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, VIPAS39, VPS33B, WDR72, XDH		
Hyperkalziurie: 16 Gene (32,3 kb)		
ADCY10, BSND, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, RRAGD, SLC12A1, SLC34A1, SLC34A3		
Hyperoxalurie: 5 Gene (6,3 kb)		
AGXT, GRHPR, HOGA1, SLC26A1, OXGR1		
Hyperglycinurie: 3 Gene (5,2 kb)		
SLC6A19, SLC6A20, SLC36A2		
Hypocitraturie: 4 Gene (11,1 kb)		
TP6V0A4, ATP6V1B1, SLC4A1, SLC4A4		
Xanthinurie: 2 Gene (6,7 kb)		
MOCOS, XDH		
Cystinurie: 2 Gene (3,5 kb)		
SLC3A1, SLC7A9		
Nierenzellkarzinom *	3 - 5 Wo	E
Gen-Panel: ID041.04, 34 Gene (76,8 kb)		
BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1		
Nephroblastom und Wilms-Tumor (WT) *	3 - 5 Wo	E
Gen-Panel: ID335.01, 30 Gene (97,7 kb)		
AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CHEK2, CTR9, DICER1, DIS3L2, FBXW7, FIBP, GPC3, GPC4, KDM3B, NSD1, NYNRIN, MLH1, MSH2, MSH6, PALB2, PMS2, POU6F2, REST, TP53, TRIM28, TRIM37, TRIP13, WT1		
Nephrokalzinose *	3 - 5 Wo	E
Gen-Panel: ID361.01, 32 Gene (56,7 kb)		
ADCY10, AGXT, ALPL, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, FAH, FAM20A, GRHPR, HNF4A, HOGA1, KCNJ1, MAGED2, OCRL, OXGR1, PHEX, RRAGD, SLC12A1, SLC34A1, SLC34A3, SLC4A1, STRADA, VIPAS39, VPS33B		
Nephronophthise (NPHP) *	3 - 5 Wo	E
Gen-Panel: ID030.02, 22 Gene (70,9 kb)		
ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423		

Erkrankung/Diagnostik	Dauer	Material
Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS) *	4 - 6 Wo	E
Gen-Panel: ID098.06		
Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS): 43 Gene (129,7 kb)		
ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, DAAM2, DGKE, EMP2, INF2, KANK2, KIRREL1, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PAX2, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, TRPC6, WT1		
Nephrotisches Syndrom (SRNS, NPHS): 30 Gene (89,3 kb)		
ARHGAP24, ARHGDIA, AVIL, COQ2, COQ6, COQ8B, DAAM2, DGKE, EMP2, KANK2, KIRREL1, LAMA5, LAMB2, MAGI2, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, WT1		
Fokal-segmentale Glomerulosklerose (FSGS): 18 Gene (49,5 kb)		
ACTN4, ANLN, APOL1, ARHGAP24, COQ2, COQ6, COL4A3, COL4A4, COL4A5, CRB2, CD2AP, INF2, LMX1B, MYO1E, PAX2, PDSS2, SMARCAL1, TRPC6		
Polyzystische Lebererkrankung (PCLD) *	3 - 5 Wo	E
Gen-Panel: ID305.02		
Polyzystische Lebererkrankung (PCLD): 16 Gene (55,7 kb)		
ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63		
Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 7 Gene (14,0 kb)		
ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B		
Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 9 Gene (41,8 kb)		
ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1		
Polyzystische Nierenerkrankung (PKD) *	3 - 5 Wo	E
Gen-Panel: ID295.03		
Polyzystische Nierenerkrankung (PKD): 9 Gene (41,8 kb)		
ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1		
Polyzystische Nierenerkrankung, autosomal-dominant (ADPKD): 7 Gene (27,4 kb)		
ALG5, DNAJB11, GANAB, IFT140, NEK8, PKD1, PKD2		
Polyzystische Nierenerkrankung, autosomal-rezessiv (ARPKD): 2 Gene (14,5 kb)		
DZIP1L, PKHD1		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) *	3 - 5 Wo	E
Gen-Panel: ID250.01		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA): 8 Gene (23,8 kb)		
CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4		
Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb)		
NR3C2, SCNN1A, SCNN1B, SCNN1G		
Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb)		
CUL3, KLHL3, WNK1, WNK4		
Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb)		
SCNN1A, SCNN1B, SCNN1G		
Renale Amyloidose *	3 - 5 Wo	E
Gen-Panel ID320.01		
Renale Amyloidose: 9 Gene (13,2 kb)		
APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TNFRSF1A, TTR		
Hereditäre systemische Amyloidose (AMYLD): 6 Gene (6,3 kb)		
APOA1, B2M, FGA, GSN, LYZ, TTR		
Periodische Fiebersyndrome mit Amyloidose: 3 Gene (6,8 kb)		
MEFV, NLRP3, TNFRSF1A		
Renale Hypodysplasie, Aplasie und Agenesie *	3 - 5 Wo	E
Gen-Panel: ID319.00, 23 Gene (72,5 kb)		
BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4		
Renale tubuläre Azidose (RTA) *	3 - 5 Wo	E
Gen-Panel: ID297.00, 9 Gene (18,5 kb)		
ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72		
Renale tubuläre Dysgenesie (RTD) *	2 - 4 Wo	E
Gen-Panel: ID316.00, 4 Gene (7,7 kb)		
ACE, AGT, AGTR1, REN		
Renale Ziliopathien, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID376.00		
Renale Ziliopathien, umfassende Diagnostik: 75 Gene (181,7 kb)		
AHI1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPGRIP1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, XPNPEP3, ZNF423		
Nephronophthise (NPHP): 17 Gene (50,3 kb)		
ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423		
Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb)		
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Senior-Loken-Syndrom (SLSN): 8 Gene (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Joubert-Syndrom (JBTS): 40 Gene (104,1 kb)		
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423		
Meckel-Syndrom (MKS): 14 Gene (36,2 kb)		
B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15		

Erkrankung/Diagnostik	Dauer	Material
Renotubuläres Fanconi-Syndrom (FRTS) *		
Gen-Panel: ID359.00, 7 Gene (10,4 kb)	3 - 5 Wo	E
CTNS, EHHADH, GATM, HNF4A, NDUFAF6, SLC2A2, SLC34A1		
Senior-Loken-Syndrom (SLSN) *		
Gen-Panel: ID029.01, 8 Gene (27,9 kb)	3 - 5 Wo	E
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Thrombotische Mikroangiopathie (TMA) *		
Gen-Panel: ID707.00, 23 Gene (44,7 kb)	3 - 5 Wo	E
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD		
Tubulointerstitielle Nierenerkrankung, autosomal-dominant (ADTKD) *		
Gen-Panel: ID296.00, 6 Gene (8,8 kb)	3 - 5 Wo	E
DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD		
Urothelkarzinom *		
Gen-Panel: ID337.00, 34 Gene (105,0 kb)	3 - 5 Wo	E
APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC		
Vesikoureteraler Reflux (VUR) *		
Gen-Panel: ID314.00, 10 Gene (33,7 kb)	3 - 5 Wo	E
DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB		
Zystische Nierenerkrankungen, umfassende Diagnostik *		
Gen-Panel: ID100.09		
Zystische Nierenerkrankungen, umfassende Diagnostik: 53 Gene (165,3 kb)		
ALG5, ALG6, ALG8, ALG9, ANKS6, BICC1, CEP83, CEP164, CEP290, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, FLCN, GANAB, GLIS2, HNF1B, IFT140, IFT172, INVS, IQCB1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, REN, RPGRIP1L, SDCCAG8, SEC61A1, SEC61B, SEC63, SLC41A1, TSC1, TSC2, TTC21B, TMEM67, UMOD, VHL, WDR19, XPNPEP3, ZNF423		
Polyzystische Nierenerkrankung (PKD): 9 Gene (41,8 kb)	4 - 6 Wo	E
ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1		
Polyzystische Lebererkrankung mit Nierenzysten (PCLD): 7 Gene (14,0 kb)		
ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B		
Autosomal-dominante tubulointerstitielle Nierenerkrankung (ADTKD): 5 Gene (7,7 kb)		
HNF1B, MUC1, REN, SEC61A1, UMOD		
Nephronophthise (NPHP): 17 Gene (50,3 kb)		
ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423		
Tuberöse Sklerose (TSC): 2 Gene (8,9 kb)		
TSC1, TSC2		
Skelett- und Knochenerkrankungen		
Adams-Oliver-Syndrom (AOS) *		
Gen-Panel: ID259.00, 6 Gene (23,2 kb)	3 - 5 Wo	E
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		
Akrozephalosyndaktylie (ACS) *		
Gen-Panel: ID311.00, 6 Gene (17,0 kb)	3 - 5 Wo	E
FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1		
Arachnodaktylie *		
Gen-Panel: ID124.00, 13 Gene (35,9 kb)	3 - 5 Wo	E
CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFBR1, TGFBR2, SMAD3		
Arthrogrypose *		
Gen-Panel: ID200.01		
Arthrogrypose: 61 Gene (209,7 kb)		
ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNM2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2		
Arthrogryposis multiplex congenita (AMC): 6 Gene (58,2 kb)	4 - 6 Wo	E
ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1		
Distale Arthrogrypose (DA): 11 Gene (40,3 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Letales kongenitales Kontraktursyndrom (LCCS): 11 Gene (31,5 kb)		
ADCY6, ADGRG6, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42		
Fetale Akinesie-Deformation-Sequenz (FADS): 4 Gene (7,6 kb)		
DOK7, MUSK, NUP88, RAPSN		
Brachydaktylie (BD) *		
Gen-Panel: ID218.02		
Brachydaktylie (BD): 21 Gene (60,1 kb)		
ADAMTS10, ADAMTS17, BMP2, BMPR1B, CHST11, CHSY1, FBN1, GDF5, HOXD13, HUWE1, IHH, LTBP2, NOG, PDE3A, PITX1, PRMT7, PTHLH, ROR2, RUNX2, TBC1D24, TRPV4	3 - 5 Wo	E
Brachydaktylie (BD), nicht-syndromal: 8 Gene (10,5 kb)		
BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PTHLH, ROR2		
Brachydaktylie (BD), syndromal: 16 Gene (52,5 kb)		
ADAMTS10, ADAMTS17, CHST11, CHSY1, FBN1, GDF5, HOXD13, HUWE1, LTBP2, NOG, PDE3A, PITX1, PRMT7, RUNX2, TBC1D24, TRPV4		
Distale Arthrogrypose (DA) *		
Gen-Panel: ID196.02, 11 Gene (40,5 kb)	3 - 5 Wo	E
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Fraser-Syndrom (FRASRS) *		
Gen-Panel: ID317.00, 3 Gene (24,8 kb)	3 - 5 Wo	E
FRAS1, FREM2, GRIP1		

Erkrankung/Diagnostik	Dauer	Material
Frontonasale Dysplasie (FND) *		
Gen-Panel ID339.00: 11 Gene (22,1 kb)	3 - 5 Wo	E
ALX1, ALX3, ALX4, ANKH, EFNB1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6		
Handfehlbildungen, umfassende Diagnostik *		
Gen-Panel: ID298.00		
Handfehlbildungen, umfassende Diagnostik: 110 Gene (295,9 kb)		
ADAMTS10, ADAMTS17, AKT3, BHLHA9, BMP2, BMPR1B, C2CD3, CACNA1C, CCND2, CCNQ, CDH3, CHST11, CHSY1, CIBAR1, CKAP2L, CPLANE1, CREBBP, DACT1, DDX59, DHCR7, DHODH, DLL4, DLX5, DOCK6, EFNB1, EFTUD2, EOGT, EP300, ESCO2, EVC2, FBLN1, FBN1, FGF10, FGF16, FGF9, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GATA6, GDF5, GDF6, GJA1, GLI1, GLI2, GLI3, GRIP1, HOXA13, HOXD13, HUWE1, IFT57, IGF2, IHH, INTU, IQCE, IRF6, KIAA0753, KIAA0825, KIF7, LMBR1, LMNA, LRP4, LTBP2, MAP3K20, MECOM, MEGF8, MYCN, NAA10, NECTIN1, NECTIN4, NOG, NOTCH1, OFD1, PAX3, PDE3A, PDE4D, PIK3CA, PIK3R2, PITX1, PRKAR1A, PRMT7, PTHLH, RAB23, RBM8A, RBPJ, RECQL4, RIPK4, ROR2, RUNX2, SALL1, SALL4, SF3B4, SMO, SMOC1, SOST, TBC1D24, TBX15, TBX3, TBX5,TCTN3, TMEM107, TP63, TRPV4, TWIST1, WDPCP, WNT10B, WNT7A, YY1AP1, ZNF141		
Brachydaktylie (BD), nicht-syndromal: 9 Gene (14,0 kb)		
BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PDE3A, PTHLH, ROR2		
Polydaktylie (PAPA, PPD), nicht-syndromal: 9 Gene (20,9 kb)		
CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141		
Syndaktylie (SDTY), nicht-syndromal: 8 Gene (18,5 kb)		
BHLHA9, FBLN1, GJA1, GLI3, HOXD13, LMBR1, LRP4, NECTIN4	4 - 6 Wo	E
Ektrodaktylie (SHFM): 7 Gene (10,6 kb)		
CDH3, DLX5, FGFR1, IGF2, TP63, WNT7A, WNT10B		
Akrozephalosyndaktylie (ACS): 6 Gene (17,0 kb)		
FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1		
Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Lakrimoaurikulodentodigitales Syndrom (LADD): 3 Gene (5,5 kb)		
FGF10, FGFR2, FGFR3		
Multiple Synostosen-Syndrom (SYNS): 6 Gene (8,3 kb)		
FGF9, GDF5, GDF6, HOXA11, MECOM, NOG		
Akrodysostosis-Syndrom (ACRDYS): 3 Gene (4,9 kb)		
PDE4D, PRKAR1A, SF3B4		
Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Herz-Hand-Syndrom *		
Gen-Panel: ID165.01, 9 Gene (23,8 kb)	3 - 5 Wo	E
DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5		
Hypophosphatasie, Hypophosphatämie und Rachitis *		
Gen-Panel: ID269.03		
Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb)		
ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR		
Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb)	3 - 5 Wo	E
CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3		
Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb)		
CYP2R1, CYP3A4, CYP27B1, VDR		
Hypophosphatasie (HPP): 1 Gen (1,6 kb)		
ALPL		
Klippel-Feil-Syndrom (KFS) *		
Gen-Panel: ID207.00, 5 Gene (12,5 kb)	3 - 5 Wo	E
GDF3, GDF6, MEOX1, MYO18B, PAX1		
Kraniosynostose (CRS) *		
Gen-Panel: ID224.02		
Kraniosynostose (CRS): 36 Gene (84,9 kb)		
ALPL, ALX4, ASXL1, CD96, CDC45, COLEC10, COLEC11, CYP26B1, EFNB1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT43, IFT122, IL11RA, MASP1, MEGF8, MSX2, P4HB, POR, PPP3CA, RAB23, RECQL4, SCARF2, SEC24D, SKI, SMAD6, TCF12, TWIST1, WDR19, WDR35, ZIC1		
Kraniosynostose (CRS), nicht-syndromal: 8 Gene (10,5 kb)		
ALX4, ERF*, IL11RA, MSX2, SMAD6, TCF12, TWIST1, ZIC1	3 - 5 Wo	E
Akrozephalosyndaktylie (ACS): 6 Gene (17,0 kb)		
FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1		
Kranioektodermale Dysplasie (CED): 4 Gene (12,1 kb)		
IFT43, IFT122, WDR19, WDR35		
Trigonozephalie (TRIGNO): 5 Gene (17,0 kb)		
ASXL1, CD96, FGFR1, FREM1, PPP3CA		
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD) *		
Gen-Panel: ID067.00		
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD): 20 Gene (68,2 kb)		
CEP120, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60	3 - 5 Wo	E
Kurzrippen-Thoraxdysplasie (Jeune-ATD, SRPS, SRTD): 18 Gene (61,3 kb)		
CEP120, DYNC2H1*, DYNC2LI1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60		
Ellis-van-Creveld-Syndrom (EVC): 4 Gene (11,5 kb)		
DYNC2L1, EVC, EVC1, WDR35		
Letales kongenitales Kontraktursyndrom (LCCS) *		
Gen-Panel: ID197.00, 12 Gene (34,6 kb)	3 - 5 Wo	E
ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42		

Erkrankung/Diagnostik	Dauer	Material
Lippen-, Kiefer- und Gaumenspalte (OFC, CLP) *	4 - 6 Wo	E
Gen-Panel: ID266.01		
Lippen-, Kiefer- und Gaumenspalte (OFC, CLP): 50 Gene (151,0 kb)		
ARHGAP29, BMP4, BRD4, C2CD3, CDH1, CHD7, CDH11, CPLANE1, CYFIP1, DDX59, DHCR7, DLX4, ESCO2, FGFR1, FGFR2, FLNB, GRHL3, HDAC8, IFT57, INTU, IRF6, KDM1A, KDM6A, KIAA0753, KMT2D, LRRC32, MEIS2, MID1, MSX1, NECTIN1, NEK1, NIPA1, NIPA2, NIPBL, OFD1, RAD21, RAB34, RIPK4, SCNM1, SLC26A2, SMC1A, SMC3, SPECC1L, TBX22, TCTN3, TGDS, TMEM107, TP63, TUBGCP5, ZRSR2		
Orofaziale Spalte, nicht-syndromal (OFC): 9 Gene (15,1 kb)		
ARHGAP29, BMP4, DLX4, GRHL3, IRF6, MSX1, NECTIN1, TBX22, TP63		
Orofaziodigitales Syndrom (OFD): 13 Gene (36,5 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TCTN3, TMEM107, ZRSR2		
Cornelia-de-Lange-Syndrom (CDLS): 6 Gene (22,9 kb)		
BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3		
Kabuki-Syndrom (KABUK): 3 Gene (23,5 kb)		
KDM1A, KDM6A, KMT2D		
Van-der-Woude-Syndrom (VWS): 2 Gene (3,3 kb)		
GRHL3, IRF6		
Mandibulofaziale Dysostose (MFD) *	3 - 5 Wo	E
Gen-Panel: ID188.01		
Mandibulofaziale Dysostose (MFD): 11 Gene (22,0 kb)		
DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D, RPS28, SF3B4, TCOF1, TSR2		
Treacher-Collins-Syndrom (TCS): 4 Gene (9,4 kb)		
POLR1B, POLR1C, POLR1D, TCOF1		
Multiple epiphysäre Dysplasie (EDM) *	3 - 5 Wo	E
Gen-Panel: ID202.02, 11 Gene (27,7 kb)		
CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CSGALNACT1, EIF2AK3, KIF7, MATN3, SLC26A2		
Multiples Pterygium-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID158.01, 8 Gene (16,8 kb)		
CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4		
Orofaziodigitales Syndrom (OFD) *	3 - 5 Wo	E
Gen-Panel: ID265.01, 14 Gene (40,2 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TBC1D32, TCTN3, TMEM107, ZRSR2		
Osteogenesis imperfecta (OI) *	3 - 5 Wo	E
Gen-Panel: ID066.02, 21 Gene (39,2 kb)		
ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1		
Osteopetrose (OPT) und ähnliche Knochenerkrankungen *	3 - 5 Wo	E
Gen-Panel ID346.01		
Osteopetrose (OPT) und ähnliche Knochenerkrankungen: 32 Gene (68,1 kb)		
AMER1, ANKH, CA2, CLCN7, CSF1R, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, LRP6, LRRK1, OSTM1, PLEKHM1, PTDSS1, PTH1R, SLC4A2, SLC29A3, SLCO2A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP		
Osteopetrose (OPTA, OPTB): 13 Gene (30,2 kb)		
CA2, CLCN7, FERMT3, LRP5, LRP6, OSTM1, PLEKHM1, SLC4A2, SLC29A3, SNX10, TCIRG1, TNFRSF11A, TNFSF11		
Syndromale Skelettdysplasie mit erhöhter Knochendichte: 19 Gene (38,0 kb)		
AMER1, ANKH, CSF1R, CTSK, DLX3, FAM20C, GJA1, HPGD, LEMD3, LRP4, LRRK1, PTDSS1, PTH1R, SLCO2A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP		
Osteoporose *	3 - 5 Wo	E
Gen-Panel: ID115.02, 15 Gene (39,0 kb)		
CALCR, COPB2, COL1A1, COL1A2, ESR1, LGR4, LRP5, NHERF1, NOTCH2, PLS3, SGMS2, SLC34A1, VDR, WNT1, WNT11		
Pierre-Robin-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID294.01, 34 Gene (73,7 kb)		
AMER1, ARCN1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, GNAI3, MYMK, MYMX, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5		
Polydaktylie, nicht-syndromale Form *	3 - 5 Wo	E
Gen-Panel: ID166.02, 9 Gene (20,9 kb)		
CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141		
Rubinstein-Taybi-Syndrom (RSTS) *	3 - 5 Wo	E
Gen-Panel: ID142.01, 3 Gene (24,3 kb)		
CREBBP, EP300, SRCAP		
Skelettdysplasie, schwere Form *	4 - 6 Wo	E
Gen-Panel: ID056.01		
Skelettdysplasie, schwere Form: 46 Gene (144,2 kb)		
AGPS, ALPL, BMPER, CANT1, CEP120, CILK1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DLL3, DYNC2H1, EBP, FAM20C, FAM111A, FGFR2, FGFR3, FLNA, FLNB, GDF5, GNPAT, GPX4, HSPG2, IFT52, IFT80, IFT81, INPPL1, KIAA0586, LBR, LIFR, NEK1, NSDHL, P3H1, PEX5, PEX7, PPIB, PTH1R, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4, WDR34, WDR35		
Achondrogenesie (ACG): 4 Gene (14,1 kb)		
COL2A1, GDF5, SLC26A2, TRIP11		
Fibrochondrogenesie (FBCG): 2 Gene (10,6 kb)		
COL11A1, COL11A2		
Thanatophore Dysplasie (TD): 2 Gene (6,9 kb)		
COL1A2, FGFR3		
Chondrodysplasie, letal: 12 Gene (24,5 kb)		
AGPS, EBP, FLNB, GDF5, GNPAT+B343, GPX4, LBR, PEX5, PEX7, PTH1R, SLC26A2, SLC35D1		
Osteogenesis imperfecta (OI), letal: 5 Gene (12,6 kb)		
CRTAP, COL1A1, COL1A2, P3H1, PPIB		
Kurzrippen-Thoraxdysplasie (SRTD), letal: 9 Gene (35,0 kb)		
CEP120, DYNC2H1, IFT52, IFT80, IFT81, KIAA0586, NEK1, WDR34, WDR35		

Erkrankung/Diagnostik	Dauer	Material
Skelettdysplasien, umfassende Diagnostik * Gen-Panel: ID356.00, 407 Gene (969,6 kb) ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF3, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARL6, ARSB, ARSL, ASXL1, ASXL2, ATP6V0A2, ATP7A, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRTAP, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GJA1, GLB1, GLI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GSC, GUSB, GZF1, HDAC8, HES7, HGSNAT, HHAT, HOXD13, HPGD, HS2ST1, HSPG2, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IL11RA, IL1RN, INPPL1, KAT6B, KDELR2, KIAA0753, KIF22, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MBTPS1, MEGF8, MEOX1, MESD, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NSD1, NSDHL, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RINT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMAD6, SMARCAL1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFBR2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UFSP2, UNC45A, VDR, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6	4 - 6 Wo	E
Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD) * Gen-Panel: ID110.01 Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD): 39 Gene (90,7 kb) ACAN, ACP5, AIFM1, B3GALT6, BGN, CFAP410, CHST3, COL2A1, COL10A1, COL11A2, COMP, DDR2, DDRGK1, EXOC6B, FN1, GPX4, KIF22, MATN3, MBTPS1, MMP13, NANS, NEPRO, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, PLCB3, POP1, RPL13, RSPRY1, SIK3, SMARCAL1, TONSL, TRAPPC2, TRIP11, TRPV4, UFSP2, WISP3 Spondyloepiphysäre Dysplasie (SED): 9 Gene (25,9 kb) ACAN, CHST3, COL2A1, COMP, MBTPS1, SMARCAL1, TRAPPC2, TRPV4, WISP3 Spondylometaphysäre Dysplasie (SMD): 11 Gene (30,2 kb) ACP5, CFAP410, COL2A1, COL10A1, FN1, GPX4, PAM16, PCYT1A, PLCB3, TRIP11, TRPV4 Spondyloepimetaphysäre Dysplasie (SEMD): 21 Gene (47,6 kb) ACAN, AIFM1, B3GALT6, BGN, COL2A1, DDR2, DDRGK1, EXOC6B, KIF22, MATN3, MMP13, NANS, NEPRO, PAPSS2, PISD, POP1, RPL13, RSPRY1, SIK3, TONSL, UFSP2	3 - 5 Wo	E
Spondylokostale Dysostose (SCDO) * Gen-Panel: ID227.00, 7 Gene (14,4 kb) DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6	3 - 5 Wo	E
3M-Syndrom * Gen-Panel: ID214.00, 3 Gene (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 Wo	E
Stoffwechselerkrankungen		
Amyloidose * Gen-Panel: ID375.00 Amyloidose: 18 Gene (25,0 kb) APP, APOA1, APOA2, APOC2, B2M, CST3, FGA, GPNMB, GSN, IL31RA, ITM2B, LYZ, MEFV, NLRP3, OSMR, PRNP, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 8 Gene (7,0 kb) APOA1, APOA2, APOC2, B2M, FGA, GSN, LYZ, TTR Primäre lokalisierte kutane Amyloidose (PLCA): 3 Gene (7,0 kb) GPNMB, OSMR, IL31RA Zerebrale Amyloidangiopathie (HCHWA): 4 Gene (4,3 kb) APP, CST3, ITM2B, PRNP	3 - 5 Wo	E
Coenzym-Q10-Mangel (COQ10D) * Gen-Panel: ID225.01, 15 Gene (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2	3 - 5 Wo	E
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen * Gen-Panel: ID706.01, 16 Gene (30,1 kb) ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1	3 - 5 Wo	E
Folatstoffwechselstörung * Gen-Panel: ID334.00, 10 Gene (18,7 kb) CBS*, FOLR1, FOLR2, FTCD, MTHFD1, MTHFR, MTR, MTRR, SLC19A1, SLC46A1	3 - 5 Wo	E
Glykogenspeicherkrankheit (GSD) * Gen-Panel: ID108.01, 29 Gene (54,1 kb) AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC1, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4	3 - 5 Wo	E
Glykosylphosphatidylinositol (GPI)-Biosynthesedefekt (GPIBD) * Gen-Panel: ID291.00 Glykosylphosphatidylinositol (GPI)-Biosynthesedefekt (GPIBD): 22 Gene (33,3 kb) GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY Multiple kongenitale Anomalien-Hypotonie-Krampfanfälle-Syndrom (MCAHS): 4 Gene (7,7 kb) PIGA, PIGN, PIGQ, PIGT Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS): 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hämochromatose (HFE) und Hämosiderose *	3 - 5 Wo	E
Gen-Panel: ID114.04, 10 Gene (14,6 kb)		
BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2		
Homocystinurie *	3 - 5 Wo	E
Gen-Panel: ID191.01, 9 Gene (15,3 kb)		
ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1		
Hyperinsulinämische Hypoglykämie (HHF) *	3 - 5 Wo	E
Gen-Panel: ID126.00, 8 Gene (16,9 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A		
Hyperkalzämie *	3 - 5 Wo	E
Gen-Panel: ID262.00, 8 Gene (14,6 kb)		
AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1		
Hyperoxalurie *	3 - 5 Wo	E
Gen-Panel: ID363.00		
Hyperoxalurie: 6 Gene (8,6 kb)		
AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6		
Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb)		
AGXT, GRHPR, HOGA1		
Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb)		
OXGR1, SLC26A1, SLC26A6		
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *	3 - 5 Wo	E
Gen-Panel: ID292.00, 6 Gene (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung *	3 - 5 Wo	E
Gen-Panel: ID280.00		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung: 44 Gene (85,9 kb)		
ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4		
Glykogenspeicherkrankheit (GSD): 24 Gene (48,4 kb)		
AGL, ALDOA. ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4		
Hyperinsulinämische Hypoglykämie (HHF): 9 Gene (18,8 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A		
Hypomagnesiämie (HOMG) *	3 - 5 Wo	E
Gen-Panel ID054.02: 14 Gene (30,9 kb)		
ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6		
Hypophosphatasie, Hypophosphatämie und Rachitis *	3 - 5 Wo	E
Gen-Panel: ID269.03		
Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb)		
ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR		
Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb)		
CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3		
Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb)		
CYP2R1, CYP3A4, CYP27B1, VDR		
Hypophosphatasie (HPP): 1 Gen (1,6 kb)		
ALPL		
Kombinierter oxidativer Phosphorylierungsmangel (COXPD) *	3 - 5 Wo	E
Gen-Panel: ID287.01, 60 Gene (80,3 kb)		
AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2		
Kongenitale Glykosylierungsstörung (CDG) *	3 - 5 Wo	E
Gen-Panel ID035.03		
Kongenitale Störung der Glykosylierung (CDG): 58 Gene (86,2 kb)		
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, GET4, MAGT1, MAN1B1, MAN2B2, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, STX5, TMEM165, TUSC3, VMA12, VMA22		
Kongenitale Glykosylierungsstörung, Typ I (CDG1): 31 Gene (43,6 kb)		
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAN2B2, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3		
Kongenitale Glykosylierungstörung, Typ II (CDG2): 27 Gene (42,7 kb)		
ATP6AP1, ATP6AP2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, GET4, MAN1B1, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, STX5, TMEM165, VMA12, VMA22		
MODY-Diabetes *	3 - 5 Wo	E
Gen-Panel: ID048.01, 14 Gene (22,9 kb)		
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1		
Metabolische Muskelerkrankungen und Rhabdomyolyse *	4 - 6 Wo	E
Gen-Panel: ID395.00, 93 Gene (205,6 kb)		
ABHD5, ACAD9, ACADM, ACADS, ACADVL, AGK, AGL, ALDOA, AMPD1, ANO5, ATP2A2, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHKB, CNTN1, COQ4, COQ8A, CPT2, CTBP1, DMD, DTNA, DNAJB6, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FKRP, FLAD1, GAA, GBE1, GMPPB, GUK1, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LIG3, LPIN1, MGME1, MLIP, MPV17, MRM2, MYH1, OBSCN, OPA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PNPLA2, POC5, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC22A5, SLC25A10, SLC25A20, SLC25A21, SLC25A4, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TK2, TAMM41, TANGO2, TRAPPC2L, TSFM, TWNK, TYMP, YARS2		

Erkrankung/Diagnostik	Dauer	Material
Metabolische Epilepsien * Gen-Panel: ID303.01 Metabolische Epilepsien: 84 Gene (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycin-Enzephalopathie (GCE): 5 Gene (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdän-Cofaktor-Defizienz (MOCOD): 3 Gene (3,7 kb) GPHN, MOCS1, MOCS2 Zerebrales Kreatinmangelsyndrom (CCDS): 3 Gene (3,9 kb) GAMT, GATM, SLC6A8 Ahornsirupkrankheit (MSUD): 5 Gene (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxy-Glutarazidurie: 3 Gene (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-Gangliosidose: 3 Gene (3,8 kb) HEXA, HEXB, GM2A Neuronale Ceroid-Lipofuszinose (CLN): 12 Gene (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 Wo	E
Mukopolysaccharidose (MPS) * Gen-Panel: ID308.00, 12 Gene (21,2 kb) ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A	3 - 5 Wo	E
Muskeldystrophie-Dystroglykanopathie (MDDG) * Gen-Panel: ID179.00 Muskeldystrophie-Dystroglykanopathie (MDDG): 15 Gene (24,0 kb) B3GALNT2, B4GAT1, DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglykanopathie (MDDGA) mit Hirn- und Augenanomalien: 14 Gene (22,8 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD*, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglykanopathie (MDDGB) mit oder ohne intellektuelle Entwicklungsstörung: 8 Gene (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2 Muskeldystrophie-Dystroglykanopathie (MDDGC), Gliedergürtelmuskeldystrophie: 11 Gene (18,5 kb) DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, POMGNT1, POMGNT2, POMK, POMT1, POMT2	3 - 5 Wo	E
Neonataler Diabetes mellitus * Panel: ID162.01 Neonataler Diabetes mellitus: 29 Gene (53,6 kb) ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57 Permanenter neonataler Diabetes mellitus (PNDM): 10 Gene (15,9 kb) ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57 Syndromaler neonataler Diabetes mellitus: 21 Gene (40,5 kb) BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 Wo	E
Neuronale Ceroid-Lipofuszinose (CLN) * Gen-Panel: ID132.01, 15 Gene (20,2 kb) ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1	3 - 5 Wo	E
Porphyrie * Gen-Panel: ID153.01, 10 Gene (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 Wo	E
Pulmonale Surfactant-Stoffwechselstörung (SMDP) * Gen-Panel: ID168.01, 6 Gene (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 Wo	E
Pyruvatdehydrogenase-Mangel (PDHD) * Gen-Panel: ID393.00, 7 Gene (10,0 kb) DLAT, DLD, LIAS, PDHA1, PDHB, PDHX, PDP1	3 - 5 Wo	E
Renale Amyloidose * Gen-Panel ID320.01 Renale Amyloidose: 9 Gene (13,2 kb) APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 6 Gene (6,3 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Periodische Fiebersyndrome mit Amyloidose: 3 Gene (6,8 kb) MEFV, NLRP3, TNFRSF1A	3 - 5 Wo	E
Speicherkrankheiten mit Herzbeteiligung * Gen-Panel: ID149.03 Speicherkrankheiten mit Herzbeteiligung: 16 Gene (20,7 kb) APOA1, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Kardiale Glykogenspeicherkrankheit (GSD): 3 Gene (5,8 kb) GAA, LAMP2, PRKAG2 Hämochromatose (HFE): 6 Gene (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Amyloidose (AMYLD): 6 Gene (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Störung der Peroxisomenbiogenese (PBD) *	3 - 5 Wo	E
Gen-Panel: ID083.01		
Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Neonatale Adrenoleukodystrophie / Infantiles Refsum-Syndrom (PBD, Typ B): 11 Gene (17,0 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26		
Heimler-Syndrom (PBD, Typ C): 2 Gene (6,8 kb)		
PEX1, PEX6		
Stoffwechselstörung mit Epilepsie im Neugeborenenalter *	3 - 5 Wo	E
Gen-Panel: ID135.00, 25 Gene (39,4 kb)		
ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO		
Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter *	3 - 5 Wo	E
Gen-Panel: ID171.00, 18 Gene (31,5 kb)		
ALDH5A1, ATP7A, BTD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1		
Stoffwechselstörung mit Epilepsie im Schul- und Jugendalter *	3 - 5 Wo	E
Gen-Panel: ID172.00, 15 Gene (28,1 kb)		
ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2		
Zellweger-Syndrom (ZWS) *	3 - 5 Wo	E
Gen-Panel: ID084.00, 14 Gene (22,3 kb)		
ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Zystische Fibrose (CF) *	3 - 5 Wo	E
Gen-Panel: ID045.00, 1 Gen (4,4 kb)		
CFTR		
3-Methylglutaconazidurie (MGCA) *	3 - 5 Wo	E
Gen-Panel: ID249.01, 16 Gene (18,9 kb)		
AGK, ATPAF2, ATP5F1D, ATP5F1E, AUH, CLPB, DNAJC19, ECHS1, HTRA2, OPA3, POLG, SERAC1, SUCLA2, TAFAZZIN, TIMM50, TMEM70		
Tumorerkrankungen		
BRCA1- und BRCA2-assoziierte Tumordisposition *	2 - 4 Wo	E
Gen-Panel: ID001.00, 2 Gene (15,8 kb)		
BRCA1, BRCA2		
Chromosomen-Instabilitätssyndrome *	4 - 6 Wo	E
Gen-Panel: ID326.01, 40 Gene (121,0 kb)		
ANAPC1, ATM, BLM, BRCA1, BRCA2, BRIP1, DDB2, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, MRE11, NBN, PALB2, PCNA, POLH, RAD50, RAD51, RAD51C, RECQL4, RFWD3, SLX4, TOP3A, UBE2T, WRN, XPA, XPC, XRCC2		
Cowden-Syndrom (CWS) *	3 - 5 Wo	E
Gen-Panel: ID075.01, 8 Gene (12,8 kb)		
AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1		
DNA-Reparatur-Defizienz-Syndrome, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID348.00, 221 Gene (507,3 kb)		
ABRAXAS1, ALKBH2, ALKBH3, ANAPC1, APEX1, APEX2, APLF, APTX, ATM, ATR, ATRIP, ATRX, BARD1, BLM, BRCA1, BRCA2, BRIP1, CCNH, CDK7, CETN2, CHAF1A, CHEK1, CHEK2, CLK2, DCLRE1A, DCLRE1B, DCLRE1, DDB1, DDB2, DMC1, DNA2, DNPH1, DNTT, DUT, EME1, EME2, ENDOV, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, EXO1, EXO5, FAAP100, FAAP20, FAAP24, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FEN1, GEN1, GTF2E2, GTF2H1, GTF2H2, GTF2H3, GTF2H4, GTF2H5, H2AX, HELQ, HERC2, HFM1, HLTF, HMCES, HUS1, LIG1, LIG3, LIG4, MAD2L2, MBD4, MDC1, MGMT, MLH1, MLH3, MMS19, MNAT1, MPG, MPLKIP, MRE11, MSH2, MSH3, MSH4, MSH5, MSH6, MUS81, MUTYH, NABP2, NBN, NEIL1, NEIL2, NEIL3, NHEJ1, NTHL1, NUDT1, NUDT15, NUDT18, OGG1, PALB2, PARG, PARK7, PARP1, PARP2, PARP3, PARPBP, PAXIP1, PCNA, PDS5B, PER1, PMS1, PMS2, PNKP, POLA1, POLB, POLD1, POLD2, POLD3, POLD4, POLE, POLE2, POLE3, POLE4, POLG, POLH, POLI, POLK, POLL, POLM, POLN, POLQ, PRIMPOL, PRKDC, PRPF19, RAD1, RAD17, RAD18, RAD23A, RAD23B, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54B, RAD54L, RAD9A, RBBP8, RDM1, RECQL, RECQL4, RECQL5, REV1, REV3L, RIF1, RFWD3, RMI1, RNF168, RNF4, RNF8, RPA1, RPA2, RPA3, RPA4, RRM2B, SEM1, SETMAR, SHLD1, SHLD2, SHLD3, SHPRH, SLX1A, SLX1B, SLX4, SMC5, SMC6, SMUG1, SPIDR, SPO11, SPRTN, SWI5, SWSAP1, TDG, TDP1, TDP2, TOP3A, TOPBP1, TP53, TP53BP1, TREX1, TREX2, UBE2A, UBE2B, UBE2N, UBE2T, UBE2V2, UNG, USP1, UVSSA, WDR48, WRN, XAB2, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, XRCC5, XRCC6, ZSWIM7		
Dyskeratosis congenita (DKC) *	3 - 5 Wo	E
Gen-Panel: ID347.01, 15 Gene (24,1 kb)		
ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53		
Erbliche Tumorerkrankungen, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID018.05, 357 Gene (796,5 kb)		
ABRAXAS1, ACD, ADA2, ADH5, AIP, AKT1, ALDH2, ALK, AMER1, ANAPC1, ANKRD26, APC, APTX, AR, ARID1A, ASXL1, ATM, ATR, ATRX, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BTK, BUB1B, CASP10, CASP8, CASR, CBL, CCND1, CDC73, CDCA7, CDH1 ,CDH23, CDK4, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CEBPA, CEP57, CHEK2, CLPB, CSF3R, CTC1, CTLA4, CTNNA1, CTR9, CTRC, CYLD, CYP3A43, DCLRE1B, DCLRE1C, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNA2, DNAJC21, DNMT3A, DNMT3B, DUT, EFL1, EGFR, EHBP1, ELAC2, ELANE, ELP1, ENOSF1, EPCAM, ERBB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ETV6, EXO1, EXT1, EXT2, EZH2, FAH, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGFR3, FH, FLCN, FOXE1, G6PC3, GALNT12, GATA1, GATA2, GDNF, GEN1, GFI1, GNAS, GPC3, GPC4, GPR101, GPR161, GREM1, GTF2E2, GTF2H5, HABP2, HAVCR2, HAX1, HCLS1, HEATR3, HELLS, HERC2, HFM1, HNF1A, HNF1B, HOXB13, HRAS, IKZF1, JAGN1, JAK2, KDM1A, KIF1B, KIT, KLHDC8B, KRAS, LAPTM5, LIG1, LIG3, LIG4, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAP2K5, MAPK1, MAX, MBD4, MC1R, MDH2, MDM4, MECOM, MEN1, MET, MGMT, MINPP1, MITF, MLH1, MLH3, MPLKIP, MRAS, MRE11, MSH2, MSH3, MSH4, MSH5, MSH6, MSR1, MTAP, MUTYH, MYD88, MYSM1, NAF1, NBN, NDUFA13, NF1, NF2, NHEJ1, NHP2, NKX2-1, NOP10, NRAS, NSD1, NTHL1, NTRK1, OGG1, PALB2, PALLD, PARK7, PARN, PAX5, PBRM1, PCNA, PDGFRA, PHOX2B, PIK3CA, PLA2G2A, PMS1, PMS2, PNKP, POLA1, POLD1, POLE, POLG, POLH, POT1, POU6F2, PRF1, PRIMPOL, PRKAR1A, PRKCD, PRKDC, PRKN, PRSS1, PTCH1, PTCH2, PTEN, PTPN11, RABL3, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54B, RAD54L, RAF1, RASA2, RB1, RBBP6, RBBP8, RBM8A, RECQL, RECQL4, REST, RET, RFC1, RFWD3, RHBDF2, RINT1, RIT1, RNASEL, RNF139, RNF168, RNF43, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RRAS2, RRM2B, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SH2B3, SH2D1A, SHOC2, SLC25A11, SLX4, SMAD4, SMAD7, SMARCA4, SMARCB1, SMARCE1, SMC5, SOS1, SOS2, SPIDR, SPINK1, SPRED1, SPRTN, SQSTM1, SRGAP1, SRP54, SRP72, STAT3, STK11, STN1, SUFU, TBXT, TDP1, TDP2, TERC, TERF2IP, TERT, TGFBR2, TINF2, TMEM127, TNFRSF11A, TOP3A, TP53, TREX1, TRIM28, TRIM37, TRIP13, TRRAP, TSC1, TSC2, TSR2, TYMS, TYR, UBE2A, UBE2T, UNC13D, UNG, USB1, USP8, USP45, UVSSA, VHL, VPS45, WAS, WRAP53, WRN, WT1, WWP1, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, ZBTB24, ZCCHC8, ZNF687, ZSWIM7		

Erkrankung/Diagnostik	Dauer	Material
Endometriumkarzinom *	3 - 5 Wo	E
Gen-Panel: ID364.00, 12 Gene (29,1 kb)		
EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, STK11, TP53		
Endometriumkarzinom, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel: ID365.00, 26 Gene (92,9 kb)		
ATM, APC, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11, TP53		
Fanconi-Anämie (FANC) *	3 - 5 Wo	E
Gen-Panel: ID043.02, 21 Gene (60,7 kb)		
BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2		
Gastrointestinaler Stromatumor (GIST) *	3 - 5 Wo	E
Gen-Panel: ID226.00, 8 Gene (19,0 kb)		
KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD		
Glioblastom (GLM) *	3 - 5 Wo	E
Gen-Panel: ID313.00, 16 Gene (61,0 kb)		
APC, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POT1, PTEN, TP53, TSC1, TSC2		
Gorlin–Syndrom (BCNS) und ähnliche Krankheitsbilder *	3 - 5 Wo	E
Gen-Panel: ID174.02, 8 Gene (25,8 kb)		
BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU		
Hypophysenadenom (PITA) *	3 - 5 Wo	E
Gen-Panel: ID387.00, 8 Gene (20,7 kb)		
AIP, CDH23, CDKN1B, GNAS, GPR101, MEN1, PRKAR1A, USP8		
Kolorektales Karzinom *	3 - 5 Wo	E
Gen-Panel: ID006.10, 24 Gene (66,3 kb)		
APC, ATM, AXIN2, BAP1, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53		
Kolorektales Karzinom mit Mikrosatelliteninstabilität (MSI) *	3 - 5 Wo	E
Gen-Panel: ID283.00, 9 Gene (25,4 kb)		
EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE		
Konstitutionelles MMR-Defizienz-Syndrom (CMMRDS, MMRCS) *	3 - 5 Wo	E
Gen-Panel: ID362.00, 5 Gene (12,7 kb)		
EPCAM, MLH1, MSH2, MSH6, PMS2		
Krebserkrankungen im Kindesalter *	4 - 6 Wo	E
Gen-Panel: ID333.00		
Krebserkrankungen im Kindesalter: 139 Gene (341,1kb)		
ACD, ALK, ANKRD26, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNAJC21, EFL1, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, GATA2, GPC3, HRAS, IKZF1, KIF1B, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PMS2, POLE, POLH, POU6F2, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RAD51, RAD51C, RAF1, RB1, RECQL4, REST, RET, RFWD3, RIT1, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, SRP72, STK11, SUFU, TERT, TMEM127, TINF2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2		
Maligne hämatologische Erkrankungen: 49 Gene (126,4 kb)		
ACD, ANKRD26, ATM, BLM, BRCA1, BRCA2, BRIP1, CEBPA, DDX41, DKC1, DNAJC21, EFL1, ELANE, ERCC4, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA2, IKZF1, MAD2L2, NBN, NHP2, NOP10, PALB2, PARN, PAX5, RAD51, RAD51C, RFWD3, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SRP72, TERT, TINF2, TP53, UBE2T, WRAP53, XRCC2		
Tumoren des Zentralnervensystems: 32 Gene (111,9 kb)		
ALK, APC, BRCA2, CDKN2A, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, KIF1B, LZTR1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PALB2, PHOX2B, PMS2, PTCH1, PTCH2, PTEN, RB1, SMARCA4, SMARCB1, SUFU, TP53, TSC1, TSC2, VHL		
Endokrine Tumoren: 15 Gene (21,2 kb)		
CDC73, CDKN1B, DLST, KIF1B, MAX, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL		
RASopathien: 18 Gene (36,6 kb)		
BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2		
Wilms-Tumor (WT): 10 Gene (29,5 kb)		
BRCA2, CDKN1C, GPC3, DIS3L2, POU6F2, TRIM28, WT1, TRIM37, CDC73, REST		
Xeroderma pigmentosum (XP): 9 Gene (19,0 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC		
MMR-Defizienz-Syndrom (MMRCS): 5 Gene (12,7 kb)		
MLH1, MSH2, PMS2, MSH6, EPCAM		
Kutanes malignes Melanom (CMM) *	3 - 5 Wo	E
Gen-Panel: ID193.01, 12 Gene (26,4 kb)		
BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3		
Lungenkarzinom *	4 - 6 Wo	E
Gen-Panel: ID260.02, 33 Gene (122,5 kb)		
ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTYH, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53		
Lynch-Syndrom (LYNCH, HNPCC) *	2 - 4 Wo	E
Gen-Panel: ID002.02, 5 Gene (12,7 kb)		
MLH1, MSH2, MSH6, PMS2, EPCAM		
Magenkarzinom *	3 - 5 Wo	E
Gen-Panel: ID090.04, 25 Gene (73,6 kb)		
APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53		
Mammakarzinom *	3 - 5 Wo	E
Gen-Panel: ID021.03, 13 Gene (41,0 kb)		
ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NTHL1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53		

Erkrankung/Diagnostik	Dauer	Material
Mamma- und Ovarialkarzinom (HBOC) *	3 - 5 Wo	E
Gen-Panel: ID003.05		
ATM, BARD1, BRIP1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NTHL1, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53		
Medulloblastom (MDB) *	3 - 5 Wo	E
Gen-Panel: ID205.02, 22 Gene (75,7 kb)		
APC, BRCA2, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTCH1, PTCH2, PTEN, SMARCB1, SMARCA4, SUFU, TP53, VHL		
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) *	4 - 6 Wo	E
Gen-Panel: ID321.01		
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML): 121 Gene (244,1 kb):		
ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8		
Akute myeloische Leukämie (AML): 12 Gene (28,4 kb)		
ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53		
Diamond-Blackfan-Anämie (DBA): 20 Gene (11,3 kb)		
GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2		
Shwachman-Diamond-Syndrom (SDS): 4 Gene (7,2 kb)		
DNAJC21, EFL1, SBDS, SRP54		
Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb)		
ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53		
Lungenfibrose und Knochenmarkinsuffizienz (PFBMFT): 6 Gene (13,0 kb)		
PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Dyskeratosis congenita (DKC): 13 Gene (21,8 kb)		
ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53		
Kongenitale Neutropenie (SCN): 10 Gene (13,9 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS		
Fanconi-Anämie (FANC): 20 Gene (60,7 kb)		
BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2		
Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb)		
MLH1, MSH2, MSH6, PMS2		
Nephroblastom und Wilms-Tumor (WT) *	3 - 5 Wo	E
Gen-Panel: ID335.01, 30 Gene (97,7 kb)		
AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CHEK2, CTR9, DICER1, DIS3L2, FBXW7, FIBP, GPC3, GPC4, KDM3B, NSD1, NYNRIN, MLH1, MSH2, MSH6, PALB2, PMS2, POU6F2, REST, TP53, TRIM28, TRIM37, TRIP13, WT1		
Neuroendokrine Neoplasie *	3 - 5 Wo	E
Gen-Panel: ID386.00, 19 Gene (29,2 kb)		
AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL		
Neurofibromatose (NF) *	3 - 5 Wo	E
Gen-Panel: ID210.00, 3 Gene (11,6 kb)		
NF1, NF2, SPRED1		
Nierenzellkarzinom *	3 - 5 Wo	E
Gen-Panel: ID041.04, 34 Gene (76,8 kb)		
BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1		
Ovarialkarzinom *	3 - 5 Wo	E
Gen-Panel: ID004.04, 14 Gene (45,5 kb)		
BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53		
Pankreaskarzinom *	3 - 5 Wo	E
Gen-Panel: ID089.04, 19 Gene (59,9 kb)		
APC, ATM, BARD1, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL, WT1		
Phäochromozytom-Paraganglíom-Syndrom (PPGL) *	3 - 5 Wo	E
Gen-Panel: ID042.03, 14 Gene (22,8 kb)		
DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL		
Plasmozytom *	3 - 5 Wo	E
Gen-Panel: ID354.01, 40 Gene (89,5 kb)		
ARID1A, ATM, BLM, BTK, CASP8, CASP10, CDKN2A, CBL, CTLA4, DDX41, EFL1, ETV6, FANCA, FAS, FASLG, HCLS1, KDM1A, KLHDC8B, KRAS, LAPTM5, MLH1, MSH2, MSH6, MYD88, NBN, NF1, NRAS, PAX5, PRF1, PMS2, POT1, PRKCD, PTPN11, RBM8A, SBDS*, SH2B3, SH2D1A, TP53, USP45, WAS		
Polyposis-Syndrom (PS, FAP) *	3 - 5 Wo	E
Gen-Panel: ID005.07, 16 Gene (41,6 kb)		
APC, AXIN2, BAP1, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11		
Prostatakarzinom *	3 - 5 Wo	E
Gen-Panel: ID140.03, 30 Gene (101,5 kb)		
AR, ATM, ATR, BAP1, BARD1, BRCA1, BRCA2, BRIP1, CYP3A43, CDH1, CHEK2, ELAC2, EPCAM, FANCM, HOXB13, MLH1, MRE11, MSR1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, RNASEL, TP53, TRRAP		
Sarkom der Weichteile und des Skelettsystems *	4 - 6 Wo	E
Gen-Panel: ID223.02, 55 Gene (155,1 kb)		
APC, ATM, ATR, ATRX, BLM, BRCA2, BUB1B, CDKN1C, CDKN2A, CEP57, CHEK2, DICER1, DKC1, EPCAM, ERCC2, EXT1, EXT2, FAH, FANCC, FH, HRAS, KIT, MEN1, MLH1, MRE11, MSH2, MSH6, MTAP, NBN, NF1, PALB2, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RPS19, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SQSTM1, SUFU, TBXT, TNFRSF11A, TP53, VHL, WAS, WRN, ZNF687		
Schilddrüsenkarzinom *	3 - 5 Wo	E
Gen-Panel: ID220.02, 26 Gene (60,5 kb)		
ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53		

Erkrankung/Diagnostik	Dauer	Material
Tuberöse Sklerose (TSC) *	2 - 4 Wo	E
Gen-Panel: ID332.00, 2 Gene (8,9 kb)		
TSC1, TSC2		
Urothelkarzinom *	3 - 5 Wo	E
Gen-Panel: ID337.00, 34 Gene (105,0 kb)		
APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC		
Xeroderma pigmentosum (XP) *	3 - 5 Wo	E
Gen-Panel: ID282.00, 10 Gene (23,5 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC		
Ziliopathien		
Bardet-Biedl-Syndrom (BBS) *	3 - 5 Wo	E
Gen-Panel: ID093.02, 21 Gene (39,0 kb)		
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Joubert-Syndrom (JBTS) *	4 - 6 Wo	E
Gen-Panel: ID028.03, 40 Gene (104,1 kb)		
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423		
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD) *	3 - 5 Wo	E
Gen-Panel: ID067.00		
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD): 20 Gene (68,2 kb)		
CEP120, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60		
Kurzrippen-Thoraxdysplasie (Jeune-ATD, SRPS, SRTD): 18 Gene (61,3 kb)		
CEP120, DYNC2H1, DYNC2LI1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60		
Ellis-van-Crefeld-Syndrom (EVC): 4 Gene (11,5 kb)		
DYNC2L1, EVC, EVC1, WDR35		
Meckel-Syndrom (MKS) *	3 - 5 Wo	E
Gen-Panel: ID032.02, 13 Gene (35,1 kb)		
B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231		
Nephronophthise (NPHP) *	3 - 5 Wo	E
Gen-Panel: ID030.02, 22 Gene (70,9 kb)		
ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423		
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *	4 - 6 Wo	E
Gen-Panel: ID085.03, 50 Gene (164,8 kb)		
BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10		
Renale Ziliopathien, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID376.00		
Renale Ziliopathien, umfassende Diagnostik: 75 Gene (181,7 kb)		
AHI1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPGRIP1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, XPNPEP3, ZNF423		
Nephronophthise (NPHP): 17 Gene (50,3 kb)		
ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423		
Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb)		
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Senior-Loken-Syndrom (SLSN): 8 Gene (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Joubert-Syndrom (JBTS): 40 Gene (104,1 kb)		
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423		
Meckel-Syndrom (MKS): 14 Gene (36,2 kb)		
B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15		
Senior-Loken-Syndrom (SLSN) *	3 - 5 Wo	E
Gen-Panel: ID029.01, 8 Gene (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		

Erkrankung/Diagnostik	Dauer	Material
Pränatale Diagnostik: Fetale Anomalien		
Fetale Anomalien *	4 - 6 Wo	E
Gen-Panel: ID850.00, 1223 Gene (3235,6 kb)		
AAAS, ABCA12, ABCC6, ABCC9, ABHD5, ABL1, ACAD9, ACADVL, ACAN, ACE, ACOX1, ACP5, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTG2, ACVR2B, ACY1, ADAMTS10, ADAMTS17, ADAMTS3, ADAMTSL2, ADAR, ADGRG1, ADGRG6, ADNP, ADSL, AFF4, AGK, AGL, AGPS, AHCY, AHDC1, AHI1, AKT1, AKT2, AKT3, ALDH18A1, ALDH1A3, ALDH3A2, ALDH7A1, ALDOA, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALX1, ALX3, ALX4, AMACR, AMER1, AMMECR1, AMPD2, AMT, ANAPC1, ANKH, ANKRD11, ANKS6, ANOS1, ANTXR1, ANTXR2, AP1S2, AP4E1, AR, ARCN1, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6V0A2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCL11A, BCOR, BCS1L, BFSP2, BGN, BHLHA9, BICD2, BIN1, BLM, BLTP1, BMP1, BMP2, BMP4, BMPER, BMPR1B, BNC2, BPNT2, BRAF, BRAT1, BRCA2, BRIP1, BRPF1, BSND, BTD, BUB1B, C2CD3, CA2, CA8, CACNA1C, CACNA1E, CACNA1G, CANT1, CASK, CASR, CBL, CC2D2A, CCBE1, CCDC103, CCDC39, CCDC40, CCDC8, CCDC88C, CCND2, CCNQ, CDAN1, CDC45, CDC6, CDH1, CDH3, CDK13, CDK5RAP2, CDK8, CDKL5, CDKN1C, CDON, CDT1, CELSR1, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFAP53, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD4, CHD7, CHKB, CHMP1A, CHRNA1, CHRNA3, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CILK1, CIT, CKAP2L, CLCN7, CLP1, CLPB, CNOT1, CNOT3, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COLEC10, COLEC11, COLQ, COQ4, COQ9, COX7B, CPLANE1, CPT2, CRADD, CRB2, CREB3L1, CREBBP, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CSF1R, CSNK2A1, CSPP1, CTC1, CTCF, CTNNB1, CTNND1, CTSA, CTSD, CTSK, CTU2, CUL4B, CUL7, CWC27, CYP11A1, CYP11B1, CYP17A1, CYP1B1, CYP21A2, CYP26B1, CYP2U1, CYP4F22, DAG1, DARS1, DCHS1, DCX, DDR2, DDX11, DDX3X, DDX59, DENND5A, DHCR24, DHCR7, DHFR, DHODH, DIAPH1, DIS3L2, DISP1, DKC1, DLL3, DLL4, DLX5, DMPK, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB11, DNAL1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK6, DOK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSP, DSTYK, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, DYRK1A, DZIP1L, EBF3, EBP, ECEL1, EDA, EDNRA, EDNRB, EED, EFNB1, EFTUD2, EHMT1, EIF2AK3, EIF2B2, EIF2B3, EIF2S3, EIF4A3, EIF5A, ELAC2, ELN, ELOVL4, EMD, EML1, EMX2, ENPP1, EOGT, EP300, EPG5, EPHB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ESCO2, ETFA, ETFB, ETFDH, EVC, EVC2, EXOC3L2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EZH2, FAH, FAM111A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAR1, FAT4, FBLN5, FBN1, FBN2, FBXL4, FGD1, FGF10, FGF3, FGF8, FGFR1, FGFR2, FGFR3, FH, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FLNC, FLT4, FLVCR2, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMD4A, FTL, FUT8, FYCO1, FZD2, G6PC3, GAA, GALC, GALE, GALK1, GALNS, GALNT2, GANAB, GATA2, GATA3, GATA4, GATA6, GBA1, GBA2, GBE1, GCDH, GDF1, GDF5, GDF6, GFAP, GFM1, GFPT1, GJA1, GJA3, GJA8, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLI1, GLI2, GLI3, GLIS3, GLUL, GMNN, GMPPB, GNAI3, GNAO1, GNAS, GNB1, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC6, GPI, GPSM2, GREB1L, GRHL3, GRIN1, GRIN2B, GRIP1, GSC, GTF2H5, GTPBP3, GUCY2C, GUSB, GZF1, H1-4, HAAO, HADHA, HADHB, HBA1, HBA2, HCCS, HCFC1, HDAC8, HES7, HESX1, HIBCH, HIVEP2, HMGA2, HNF1B, HNF4A, HNRNPK, HOXA1, HOXA13, HOXD13, HPSE2, HR, HRAS, HSD17B3, HSD17B4, HSF4, HSPD1, HSPG2, HUWE1, HYCC1, HYLS1, IARS1, IDH1, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGF2, IGHMBP2, IHH, IKBKG, IL11RA, IL1RAPL1, INPP5E, INPPL1, INSR, INTU, INVS, IQCB1, IRF6, ITGA3, ITGA6, ITGA8, ITGB4, JAG1, KANSL1, KAT6A, KAT6B, KATNB1, KCNJ1, KCNJ2, KCTD1, KDM5C, KDM6A, KIAA0586, KIAA0753, KIF11, KIF14, KIF1A, KIF1B, KIF22, KIF2A, KIF5C, KIF7, KLF1, KLHL40, KLHL41, KLHL7, KMT2A, KMT2C, KMT2D, KNL1, KRAS, KRIT1, KYNU, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LARGE1, LARP7, LBR, LFNG, LGI4, LHX3, LHX4, LIFR, LIG4, LIPA, LMBR1, LMBRD1, LMNA, LMNB1, LMNB2, LMOD3, LMX1B, LONP1, LRP2, LRP4, LRP5, LRRC56, LTBP3, LTBP4, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAGEL2, MAP2K1, MAP2K2, MAP3K1, MAP3K20, MAP3K7, MAPRE2, MASP1, MATN3, MBTPS2, MCOLN1, MCPH1, MED12, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESD, MESP2, MFRP, MFSD2A, MGP, MID1, MKKS, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MMP13, MMP21, MN1, MNX1, MOCS1, MOCS2, MOGS, MPDU1, MPLKIP, MRAS, MRPS22, MSL3, MSMO1, MST01, MSX1, MSX2, MTM1, MTO1, MTOR, MTRFR, MUSK, MYBPC1, MYCN, MYH10, MYH11, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL1, MYMK, MYO18B, MYO9A, MYOCD, MYPN, MYRF, MYT1, NAA10, NACC1, NADSYN1, NAGA, NALCN, NANS, NBAS, NBN, NDE1, NDP, NDUFAF5, NEB, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEK9, NEU1, NF1, NFIX, NHEJ1, NHS, NIPAL4, NIPBL, NKX2-5, NKX3-2, NODAL, NOG, NOTCH1, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPR2, NR0B1, NR2F2, NR5A1, NRAS, NSD1, NSDHL, NUBPL, NUP107, NXN, OBSL1, OCLN, OCRL, ODAD1, ODAD2, ODAD3, OFD1, OPHN1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTX2, P3H1, P4HB, PAFAH1B1, PAK3, PALB2, PAPSS2, PARN, PAX2, PAX3, PAX6, PAX7, PAX8, PBX1, PCGF2, PCNT, PCYT1A, PDCD10, PDE4D, PDGFRB, PDHA1, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAP2, PGAP3, PGM1, PGM3, PHF6, PHF8, PHGDH, PHIP, PHOX2B, PIBF1, PIEZO1, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGT, PIGV, PIK3C2A, PIK3CA, PIK3R1, PIK3R2, PITX1, PITX2, PITX3, PKD1, PKD1L1, PKD2, PKHD1, PKLR, PLAG1, PLG, PLK4, PLOD1, PLOD2, PMM2, PNKP, PNPLA1, POC1A, POGZ, POLE, POLG2, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POP1, POR, PORCN, POU1F1, PPIB, PPP1CB, PPP2R1A, PPP2R5D, PQBP1, PRG4, PRIM1, PRKAG2, PRKAR1A, PRKD1, PRMT7, PRRX1, PRSS56, PRUNE1, PSAP, PSAT1, PSPH, PTCH1, PTDSS1, PTF1A, PTH1R, PTHLH, PTPN11, PTPN14, PTS, PUF60, PYCR1, PYGM, QRICH1, RAB18, RAB23, RAB33B, RAB3GAP1, RAB3GAP2, RAC1, RAD21, RAF1, RAI1, RAPSN, RARB, RARS2, RASA1, RAX, RBBP8, RBM10, RBM8A, RBPJ, RECQL4, RELN, REN, RERE, RET, RFT1, RFX6, RIPK4, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU4ATAC, ROBO1, ROBO3, ROGDI, ROR2, RPGRIP1L, RPL10, RPL11, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7, RRAS2, RRM2B, RSPH4A, RSPH9, RTEL1, RTTN, RUNX2, RXYLT1, RYR1, SALL1, SALL4, SAMD9, SAMHD1, SATB2, SBDS, SC5D, SCARF2, SCLT1, SCN1A, SCN2A, SCN4A, SCO2, SDCCAG8, SDR9C7, SEC23B, SEC24D, SEPSECS, SERPINF1, SERPINH1, SETBP1, SETD5, SF3B4, SGCG, SGPL1, SH3PXD2B, SHANK3, SHH, SHOC2, SHOX, SIK3, SIL1, SIX3, SIX5, SIX6, SKI, SKIC3, SLC10A7, SLC12A1, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC18A3, SLC25A19, SLC25A20, SLC25A24, SLC25A38, SLC26A2, SLC26A3, SLC27A4, SLC29A3, SLC2A10, SLC33A1, SLC35A2, SLC35C1, SLC35D1, SLC39A8, SLC5A7, SLC6A9, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC1, SMARCE1, SMC1A, SMC3, SMCHD1, SMG9, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNORD118, SNRPB, SNX10, SNX14, SON, SOS1, SOS2, SOST, SOX10, SOX17, SOX18, SOX2, SOX3, SOX6, SOX9, SP7, SPAG1, SPARC, SPATA5, SPECC1L, SPEG, SPG11, SPRED1, SRCAP, SRD5A2, SRD5A3, SRY, ST14, STAC3, STAG2, STAMBP, STAR, STIL, STRA6, STRADA, SUCLG1, SUFU, SULT2B1, SUMF1, SUZ12, TAB2, TAF1, TAFAZZIN, TALDO1, TAPT1, TBC1D20, TBC1D23, TBC1D24, TBC1D32, TBCD, TBCE, TBCK, TBL1XR1, TBX1, TBX15, TBX18, TBX20, TBX3, TBX4, TBX5, TBX6, TCF12, TCF4, TCIRG1, TCOF1, TCTN1, TCTN2, TCTN3, TELO2, TENM3, TENT5A, TFAP2A, TFAP2B, TGDS, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1, THOC6, THRA, TINF2, TMCO1, TMEM107, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM38B, TMEM67, TMEM94, TMEM98, TMX2, TNNI2, TNNT1, TNNT3, TOE1, TOP3A, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRAF7, TRAIP, TRAP1, TRAPPC12, TRAPPC9, TREX1, TRIM37, TRIP11, TRIP12, TRIP4, TRMT10A, TRPS1, TRPV4, TRPV6, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TTC21B, TTC7A, TTC8, TTN, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TWIST2, TXNDC15, TXNL4A, UBA1, UBE2T, UBE3B, UBR1, UMPS, UROS, USP18, USP9X, VAMP1, VEGFC, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDPCP, WDR19, WDR26, WDR35, WDR62, WDR73, WDR81, WNT1, WNT10B, WNT5A, WNT7A, WRAP53, WT1, XRCC4, XYLT1, XYLT2, YY1, ZBTB18, ZBTB20, ZC4H2, ZEB2, ZFP57, ZIC1, ZIC2, ZIC3, ZMPSTE24, ZMYND10, ZSWIM6		

Erkrankung/Diagnostik	Dauer	Material
Präventionsdiagnostik: Carrier-Screening		
Carrier-Screening * Gen-Panel: ID350.02, 755 Gene (1852,9 kb) AAAS, ABCA12, ABCA3, ABCA4, ABCB4, ABCB11, ABCC6, ABCC8, ABCD1, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACOX1, ACSF3, ADA, ADAMTS13, ADAMTS2, ADGRG1, ADGRV1, ADSL, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMPD1, AMT, ANK1, ANO10, ANTXR2, AP1S2, AP3B1, APTX, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS9, BCHE, BCKDHA, BCKDHB, BCOR, BCS1L, BLM, BLOC1S6, BRIP1, BRWD3, BSND, BTD, BTK, CA2, CANT1, CAPN3, CASK, CASQ2, CASR, CBS, CC2D1A, CC2D2A, CCDC88C, CD19, CD3D, CD3E, CD3G, CD40LG, CDH23, CDKL5, CEP290, CEP41, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNG, CIITA, CLCN1, CLCN5, CLCN7, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COG7, COG8, COL11A2, COL17A1, COL1A2, COL27A1, COL4A3, COL4A4, COL4A5, COL6A3, COL7A1, COQ2, COQ8A, COQ9, COX10, COX15, CPS1, CPT1A, CPT2, CRB1, CRLF1, CRTAP, CSTB, CTNS, CTSC, CTSD, CTSK, CUL4B, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, D2HGDH, DBT, DCLRE1C, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DHDDS, DKC1, DLD, DLG3, DLL3, DMD, DMP1, DNAH5, DNAI1, DNAI2, DNMT3B, DOCK8, DOK7, DOLK, DPAGT1, DPM1, DPYD, DSP, DUOX2, DUOXA2, DYNC2H1, DYSF, EDA, EDAR, EDN3, EDNRB, EFEMP2, EGR2, EIF2AK3, EIF2B5, ELP1, EMD, ENPP1, EPB42, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, F11, F2, F5, F8, F9, FAH, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCG, FASTKD2, FBLN5, FBP1, FERMT3, FGA, FGB, FGD4, FGG, FH, FKRP, FKTN, FLNA, FMR1, FMO3, FOLR1, FOXN1, FOXP3, FRAS1, FREM2, FTSJ1, FUCA1, G6PC1, G6PC3, G6PD, GAA, GBA1, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GATA1, GATM, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GDF5, GDI1, GFM1, GJA1, GJB1, GJB2, GJB3, GJB6, GJC2, GLA, GLB1, GLDC, GLE1, GNE, GNPTAB, GNPTG, GNRHR, GNS, GP1BA, GP1BB, GP9, GPC3, GRHPR, GRIK2, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HESX1, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HMOX1, HOGA1, HPD, HPRT1, HPS1, HPS3, HPS4, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HUWE1, HSD3B2, HSD3B7, HSPG2, HYAL1, HYCC1, HYLS1, ICOS, IDS, IDUA, IFNGR1, IFNGR2, IFT80, IGBP1, IGF1, IGHMBP2, IGSF1, IL12B, IL12RB1, IL1RAPL1, IL1RN, IL2RG, IL7R, INPP5E, INS, INSR, INVS, IQCB1, ITGA6, ITGB3, ITGB4, IVD, IYD, JAK3, KCNJ1, KCNJ11, KCNQ1, KCTD7, KDM5C, KRT18, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LARGE1, LBR, LCA5, LDLR, LDLRAP1, LHCGR, LHX3, LIFR, LIG4, LIPA, LIPH, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MBTPS2, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MECP2, MED12, MED17, MEFV, MESP2, MFSD8, MGAT2, MID1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPL, MPV17, MPZ, MRE11, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTTP, MUTYH, MVK, MYD88, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFA1, NDUFAF2, NDUFAF4, NDUFAF5, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NEU1, NEUROG3, NHEJ1, NHLRC1, NHS, NLGN4X, NLRP7, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR2E3, NSUN2, NTRK1, NUP62, OAT, OCA2, OCRL, OFD1, OPA3, OPHN1, ORAI1, OSTM1, OTC, OXCT1, P3H1, PAH, PAK3, PANK2, PC, PCBD1, PCCA, PCCB, PCDH15, PCDH19, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX12, PEX13, PEX2, PEX5, PEX6, PEX7, PFKM, PHGDH, PHKB, PIGN, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POLH, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRSS12, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RDH12, RELN, REN, RFT1, RLBP1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPE65, RPGR, RPGRIP1L, RPL10, RPS6KA3, RRM2B, RS1, RTEL1, SACS, SAMD9, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO1, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SHROOM4, SIL1, SKIC3, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A1, SLC4A11, SLC5A2, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMARCAL1, SMN1, SMPD1, SNAP29, SOX3, SP110, SPR, SRD5A2, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STIM1, STRA6, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYP, TAFAZZIN, TAT, TBCE, TCIRG1, TECPR2, TF, TFR2, TG, TGM1, TH, TIMM8A, TK2, TMC1, TMEM216, TMEM67, TNFRSF11B, TNNT1, TPO, TPP1, TRAPPC9, TREX1, TRHR, TRIM32, TRIM37, TRMU, TSEN54, TSFM, TSHB, TSHR, TSPYL1, TTN, TTPA, TUFM, TUSC3, TWNK, TYMP, TYR, TYRP1, UBA1, UBE2A, UBR1, UGT1A1, UNC13D, UPB1, UPF3B, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VDR, VIPAS39, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, VWF, WAS, WNT10A, WNT3, WNT7A, WRN, XIAP, XPA, XPC, ZDHHC9, ZFYVE26, ZIC3, ZMPSTE24, ZNF469, ZNF711	4 - 6 Wo	E
Whole Exome Sequencing (WES)		
WES-Solo-Exom * (Index-Patient)	1 - 3 Mo	E
WES-Trio-Exom * (Index-Patient und Eltern)	1 - 3 Mo	E
Molekulare Karyotypisierung (Array-CNV-Analyse)		
Untersuchungsmaterial: 3 - 5 ml EDTA-Blut (E), Abortgewebe (A) Änderung des EBM für humangenetische Leistungen zum 01. Juli 2016: Vor einer Array-Analyse (hochauflösende molekulare Karyotypisierung) muss eine konventionelle Chromosomenanalyse erfolgt sein. Bitte fügen Sie der Array-CGH-Anforderung die anamnestische Angabe (Chromosomenanalyse) oder ggf. Na-Heparinblut zur Durchführung einer Chromosomenanalyse bei. *Hinweis: Die Chromosomenanalyse wird in Kooperation in unserem Partnerlabor durchgeführt.* Die Untersuchung auf Mikrodeletionen und -duplikationen kann mit der GOP 11508 nur berechnet werden, wenn die klinische Fragestellung aufgrund der Analyseergebnisse der GOP 11502 (postnatale Chromosomenanalyse) nicht vollständig beantwortet werden konnte.		
SNP-Array (Nachweis quantitativer genomischer Veränderungen) * *Hinweis: Eilige Probeneingänge und spezifische Befundkonstellationen können ggf. auch über die CNV-Analyse der NGS-Pipeline analysiert werden.*	4 - 8 Wo	E, A
Hinweise		
***** = akkreditiertes Verfahren		
co = in Kooperation		
S = Stufendiagnostik		

Einzelgen-Diagnostik

Untersuchungsmaterial

Untersuchungsmaterial:
3-5 ml EDTA-Blut (E), Fruchtwasser (FW), Chorionzotten (C), Abortgewebe (A), Tumorgewebe (T),
2-3 Wangenschleimhautabstriche (WA), Fibroblastenkultur (F) des betroffenen Gewebes

Ein aktuelles Gesamtverzeichnis unserer Leistungen und aktuelle Einsenderinformationen finden Sie unter **www.zhma.de**

Bitte beachten Sie auch unser **"Handbuch der Primärprobengewinnung".**

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
A					
Aarskog-Scott-Syndrom (AAS)	P, M	305400	FGD1	2-3 Wo	E
Abetalipoproteinämie (ABL)	P	200100	MTTP	2-3 Wo	E
Absence-Epilepsie des Kindesalters (ECA) ECA2, ECA4, ECA6	P	607681 611136 611942	GABRA1, GABRA2, CACNA1H	2-4 Wo	E
Achondrogenesie (ACG)	P, M	600972	SLC26A2, COL2A1	2-4 Wo	E
Achondroplasie *, S (ACH)	P	100800	FGFR3	2-3 Wo	E, FW, C
Acyl-CoA-Dehydrogenase-Mangel ACADSD, ACADMD, ACADVLD	P, M	201470 201450 201475	ACADS, ACADM, ACADVL	2-3 Wo	E
Adipositas *	P, M	601665 614962 614963 602025 618406	LEP, LEPR, POMC, MC4R, MC3R	2-4 Wo	E
Adrenale Hypoplasie, kongenitale (AHC)	P	300200	NR0B1 (DAX1)	2-3 Wo	E
Adrenale Hyperplasie * Typ II, Typ III, Typ IV, Typ V	P, M	201810 201910 202010 202110	HSD3B2, CYP21A2, CYP11B1, CYP17A1	2-3 Wo	E, FW, C
Adrenogenitales Syndrom (AGS) durch 21-Hydroxylase-Mangel *	P, M	201910	CYP21A2	2-3 Wo	E, FW, C
Adrenoleukodystrophie (ALD) *	P, M	300100	ABCD1	2-4 Wo	E, FW, C
Agammaglobulinämie (AGM) * AGMX1, AGM7	P, M	300755 615214	BTK, PIK3R1	2-3 Wo	E
Aicardi-Goutières-Syndrom (AGS) AGS1, AGS2, AGS3, AGS4, AGS5, AGS6, AGS7	P, M	225750 610181 610329 610333 612952	TREX1, RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1, ADAR, IFIH1	2-4 Wo	E
Akromikrische Dysplasie (ACMICD) *	P, M	102370	FBN1	3-6 Wo	E
Alagille-Syndrom (ALGS) ALGS1, ALGS2	P, M	118450 610205	JAG1, NOTCH2	3-4 Wo	E
Albinismus, okulokutaner (OCA) OCA1A, OCA1B, OCA2	P	203100 606952 203200	TYR, OCA2	2-3 Wo	E
Alpha-1-Antitrypsin-Mangel (A1ATD)	P	107400	SERPINA1	2-3 Wo	E
Alpha-Thalassämie/Mentale Retardierung-Syndrom, X-chromosomal (ATRX)	P, M	301040	ATRX	2-4 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Alport-Syndrom (ATS) * ATS (X-chromosomal), ATS (autosomal-dominant), ATS (autosomal-rezessiv)	P, M	301050 203780 104200	COL4A5, COL4A3, COL4A4	4-6 Wo	E, FW, C
Alström-Syndrom (ALMS)	P	203800	ALMS1	2-3 Wo	E
Alveolar-kapilläre Dysplasie mit Misalignment der Lungengefäße	P, M	265380	FOXF1	2-3 Wo	E
Alzheimer-Krankheit 2 (AD2)	P	104310	APOE	2-4 Wo	E
Amyloidose	P	105200	APOA1	2-3 Wo	E
Andersen kardiodysrhythmische periodische Paralyse	P, M	170390	KCNJ2	2-3 Wo	E
Androgen-Insensitivität, partielle (PAIS) *	P, M	312300	AR	2-4 Wo	E
Androgeninsensitivitäts-Syndrom (AIS) *	P, M	300068	AR	2-4 Wo	E
Angelman-Syndrom (AS) *, S	P, M, F	105830	ANCR, UBEA3	2-4 Wo	E, FW, C,
Angioödem, hereditäres (HAE)	P, M	106100	C1NH (SERPING1)	2-3 Wo	E
Aniridie	P, M	106210	PAX6	2-3 Wo	E
Aortenklappenerkrankung (AOVD) AOVD1, AOVD1	P	109730 614823	NOTCH1, SMAD6	2-4 Wo	E
Apert-Syndrom *, S	P, M	101200	FGFR2	2-3 Wo	E, FW, C
Aplastische Anämie	P	609135	NBN (NBS1), SBDS	2-3 Wo	E
Apolipoprotein-A-II-Mangel	P	107670	APOA2	2-3 Wo	E
Argininbernsteinsäure-Krankheit *	P	207900	ASL	2-3 Wo	E
Arthrogrypose, distale (DA) DA2B, DA3, DA5, DA9	P	601680 114300 108145 121050	TNNI2, PIEZO2, FBN2	2-5 Wo	E
Aromatische-L-Aminosäure-Decarboxylase-Mangel (AADC-Defizienz)	P	608643	DDC	2-3 Wo	E, FW, C
Arrhythmogene rechtsventrikuläre Dysplasie, familiäre (ARVD) ARVD1, ARVD2, ARVD5, ARVD8, ARVD9, ARVD10, ARVD11, ARVD12	P	107970 600996 604400 607450 609040 610193 610476 611528	TGFB3, RYR2, TMEM43, PKP2, DSG2, DSP, DSC2, JUP	3-6 Wo	E
Ataxia teleangiectatica (AT)	P, M	208900	ATM	2-3 Wo	E
Atelosteogenesis, Typ II (AO2)	P	256050	SLC26A2	2-3 Wo	E, FW, C
Atriumseptumdefekt (ASD) * ASD2, ASD3, ASD4, ASD5, ASD7	P	607941 614089 611363 612794 108900	GATA4, MYH6, TBX20, ACTC1, NKX2-5	2-4 Wo	E
Axenfeld-Rieger-Syndrom (RIEG) RIEG1, RIEG3	P, M	180500 602482	PITX2, FOXC1	2-3 Wo	E
B					
Bannayan-Riley-Ruvalcaba-Syndrom (BRRS) *	P, M	158350	PTEN	2-3 Wo	E
Bardet-Biedl-Syndrom (BBS) * BBS1, BBS2, BBS6, BBBS8, BBS9, BBBS10, BBS12, BBS13, BBS14, BBS15	P	209900 615981 605231 615985 615986 615987 615989 615990 615991 615992	BBS1, BBS2, MKKS, TTC8, PTHB1, BBS10, BBS12, MKS1, CEP290, WDPCP	2-4 Wo	E
Barth-Syndrom (BTHS)	P	302060	TAZ	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Bartter-Syndrom (BARTS) BARTS1, BARTS2, BARTS3, BARTS4A	P, M	601678 241200 607364 602522	SLC12A1, KCNJ1, CLCNKB, BSND	2-4 Wo	E
Basalzellnävus-Syndrom (BCNS)	P, M	109400 607035	PTCH1, PTCH2, SUFU	2-4 Wo	E
Beare-Stevenson-Cutis gyrata-Syndrom (BSTVS) *	P, M	123790	FGFR2	2-3 Wo	E
Beckwith-Wiedemann-Syndrom *, S	P, M, F	130650	11p15-BWCR, CDKN1C, NSD1	2-4 Wo	E
Benigne Krampfanfälle, familiäre infantile (BFIS) BFIS2, BFIS3	P	605751 607745	PRRT2, SCN2A	2-3 Wo	E
Benigne Krampfanfälle, familiäre neonatale (BFNS) BFNS1, BFNS2	P, M	121200 121201	KCNQ2, KCNQ3	2-3 Wo	E
Bernard-Soulier-Syndrom (BSS) BSS Typ A1, BSS Typ A2, BSS Typ B, BSS Typ C	P	231200 153670	GP1BA, GP1BB, GP9	2-4 Wo	E
Beta-Thalassämie *	P, M	613985	HBB	2-3 Wo	E
Beta-Thalassämie, dominant mit Einschlusskörpern *	P, M	60902	HBB	2-3 Wo	E
Biotinidase-Mangel *	P	253260	BTD	2-4 Wo	E
Birt-Hogg-Dubé-Syndrom (BHD)	P	135150	FLNC	2-3 Wo	E
Blepharophimose, Ptosis und Epicanthus inversus (BPES)	P, M	110100	FOXL2	2-3 Wo	E
Boucher-Neuhäuser-Syndrom (BNHS)	P	215470	PNPLA6	2-3 Wo	E
Brachydaktylie, Typ B1 (BDB1)	P, M	113000	ROR2	2-3 Wo	E
Brugada-Syndrom (BRGDA) * BRGDA1, BRGDA2, BRGDA3, BRGDA4, BRGDA5, BRGDA6, BRGDA7, BRGDA8, BRGDA9, BRGDA	P, M	601144 611777 611875 611876 612838 613119 613120 613123 616399	SCN5A, GPD1L, CACNA1C, CACNB2, SCN1B, KCNE3, SCN3B, HCN4, KCND3, KCNJ8	2-4 Wo	E
Prädisposition für Brustkrebs *, S Brustkrebs, Brustkrebs early-onset, Brustkrebs lobulärer, Prädisposition für männlichen Brustkrebs, Prädisposition für Brust- und Ovarialkrebs 1/2/3/4 Es erfolgt eine Stufendiagnostik gemäß EBM Kapitel 11.4.2.	P, M	114480	BRCA1, BRCA2, RAD51C, PALB2, CHEK2, RAD51D, TP53, BRIP1, CDH1, ATM, HOXB13	2-4 Wo	E
C					
CADASIL CADASIL1, CADASIL2	P, M	125310 616779	NOTCH3, HTRA1	2-4 Wo	E
Caffey-Krankheit *	P	114000	COL1A1	2-3 Wo	E
CARASIL	P	600142	HTRA1	2-3 Wo	E
Central-Core-Myopathie (CCD) S	S	117000	RYR1	3-6 Wo	E
Ceroid-Lipofuszinose, neuronale (CLN) CLN1, CLN2, CLN3, CLN5, CLN10	P, M	256730 204500 204200 256731 610127	PPT1, TPP1, CLN3, CLN5, CTSD	2-4 Wo	E, FW, C
CETP-Mangel	P	143470	CETP	2-4 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Charcot-Marie-Tooth-Krankheit, axonal, Typ 2 (CMT2) * CMT2A2A, CMT2A2B, CMT2B1, CMT2C, CMT2E, CMT2I, CMT2J, CMT2K, CMT2M, CMT2S	P, M	609260 617087 605588 606071 607684 607677 607736 607831 606482 606071	MFN2, LMNA, TRPV4, NEFL, MPZ, GDAP1, DNM2, IGHMBP2	2-4 Wo	E
Charcot-Marie-Tooth-Krankheit, demyelinisierend, Typ 1 (CMT1) * CMT1A, CMT1B, CMT1C, CMT1D, CMT1E, CMT1F	P, M	118220 118200 601098 607678 118300 607734	PMP22, MPZ, LITAF, EGR2, NEFL	2-4 Wo	E
Charcot-Marie-Tooth-Krankheit (CMT), intermediärer Typ * CMTRIA, CMTDIB, CMTDIC, CMTRID, CMTRIE	P, M	608340 606482 608323 607791 614455	GDAP1, DNM2, YARS, MPZ, INF2	2-4 Wo	E
Charcot-Marie-Tooth-Krankheit, autosomal-rezessiv, Typ 4 (CMT4)* CMT4A, CMT4C, CMT4E, CMT4K	P, M	214400 601596 605253 616684	GDAP1, SH3TC2, MPZ, EGR2, SURF1	2-4 Wo	E
Charcot-Marie-Tooth-Krankheit, X-chrom.-dominant, 1 (CMTX1) *	P	302800	GJB1	2-3 Wo	E
CHARGE-Syndrom	P, M	214800	CHD7	2-3 Wo	E
Chilblain-Lupus (CHBL) CHBL1, CHBL2	P, M	610448 614415	TREX1, SAMHD1	2-4 Wo	E
Cholesterinester-Speicherkrankheit (CESD)	P	278000	LIPA	2-3 Wo	E
CINCA-Syndrom	P	607115	NLRP3	2-3 Wo	E
CLOVE-Syndrom *	P	612918	PIK3CA	2-3 Wo	E
COACH-Syndrom	P	216360	TMEM67, RPGRIP1L, CC2D2A	2-4 Wo	E, FW, C
Coffin-Lowry-Syndrom (CLS)	P, M	303600	RPS6KA3	2-4 Wo	E
Coffin-Siris-Syndrom (CSS) CSS1, CSS3	P, M	135900 614608	ARID1B, SMARCB1	2-4 Wo	E
Cohen-Syndrom (COH1)	P, M	216550	VPS13B	3-5 Wo	E
Cornelia-de-Lange-Syndrom (CDLS) CDLS1, CDLS2, CDLS3, CDLS5	P, M	122470 300590 610759 300882	NIPBL, SMC1A, SMC3, HDAC8	2-4 Wo	E
Corpus-callosum-Agenesie mit abnormalen Genitalien	P, M	300004	ARX	2-3 Wo	E
Costello-Syndrom (CSTLO)	P, M	218040	HRAS	2-3 Wo	E
Cowden-Syndrom (CWS) * CWS1, CWS5, CWS6, CWS7	P, M	158350 615108 615109 616858	PTEN, PIK3CA, AKT1, SEC23B	2-4 Wo	E
Cri-du-Chat-Syndrom	M	123450	5p-Chromosomenregion	2-3 Wo	E, F, C
Crigler-Najjar-Syndrom * Typ 1, Typ 2	P	218800 606785	UGT1A1	2-3 Wo	E
Crouzon-Syndrom *, S	P, M	123500	FGFR2	2-3 Wo	E, FW, C
Crouzon-Syndrom mit Acanthosis nigricans (CAN) *	P	612247	FGFR3	2-3 Wo	E
Cutis laxa, autosomal-rezessiv, Typ IC (ARCL1C)	P	613177	LTBP4	2-3 Wo	E
Cystinurie	P, M	220100	SLC3A1, SLC7A9	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
D					
Danon-Krankheit	P	300257	LAMP2	2-3 Wo	E
Denys-Drash-Syndrom (DDS)	P, M	194080	WT1	2-3 Wo	E
Diabetes insipidus, nephrogener (NDI) *, S NDI Typ 1, NDI Typ 2	P	304800 125800	AVPR2, AQP2	2-3 Wo	E
Diabetes mellitus, permanenter neonataler (PNDM) *	P	606176	ABCC8, KCNJ11, GCK, INS	2-4 Wo	E
Diabetes mellitus, transienter neonataler * TNDM2, TNDM3	P	610374 610582	ABCC8, KCNJ11	2-4 Wo	E
Diastrophische Dysplasie (DTD)	P	222600	SLC26A2	2-3 Wo	E
DiGeorge-Syndrom (DGS) S	P, M	188400 611867	22q11.2-DGCR, TBX1	2-3 Wo	E, FW
Dyggve-Melchior-Clausen-Krankheit (DMC) *	P	223800	DYM	2-3 Wo	E, FW, C
Dysgenesie des vorderen Augensegmentes (ASGD) ASGD2, ASGD3, ASGD4, ASGD5, ASGD6	P, M	610256 601631 137600 604229 617315	FOXE3, FOXC1, PITX, PAX6, CYP1B1	2-4 Wo	E
Dystonie (DYT) DYT5, DYT5b, DYT6, DYT9, DYT18, DYT24, DYT25	P, M	128230 605407 602629 615034 615073	GCH1, TH, THAP1, SLC12A1, ANO3, GNAL	2-4 Wo	E
Dilatative Kardiomyopathie, X-chromosomal (XLCM) *	P, M	302045 302060	DMD, TAZ	3-4 Wo	E
Dilatative Kardiomyopathie (DCM, CMD) * CMD1A, CMD1D, CMD1E, CMD1I, CMD1G, CMD1M, CMD1R, CMD1S, CMD1W, CMD1X, CMD1Y, CMD1Z, CMD1AA, CMD1BB, CMD1CC, CMD1DD, CMD1EE, CMD1FF, CMD1GG, CMD1HH, CMD1LL, CMD1KK, CMD1LL, CMD1MM, CMD1NN, CMD2A, CMD3B	P, M	115200 601494 601154 604145 604765 607482 613424 613426 611407 611615 611878 611879 612158 612877 613172 613122 613252 613286 613642 613881 615248 615373 615396 615916 611880 302045	LMNA, TNNT2 , SCN5A, TTN, DES, CSRP3, ACTC1, MYH7, VCL, FKTN, TPM1, TNNC1, ACTN2, DSG2, NEXN, RBM20, MYH6, TNNI3, SDHA, BAG3, MYPN, PRDM16, MYBPC3, RAF1, DMD	2-6 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
E					
Ehlers-Danlos-Syndrom (EDS) *, S EDS1, EDSCL1, EDS2, EDSCL2, EDS4, EDSVASC, EDS6, EDSKSCL1, EDS7A, EDSARTH1, EDS7B, EDSARTH2, EDS7C, EDSDERMS, EDS8, EDSPD2, EDSSPD2, EDSSPD3, EDSCLL1, EDSCLL2, EDSCV	P, M	130000 130010 130050 225400 130060 617821 225410 617174 615349 612350 606408 618000 225320	COL5A1, COL5A2, COL1A1, COL3A1, PLOD1, COL1A2, CIS, B3GALT6, SLC39A13, TNXB, AEBP1, ADAMTS2	2-5 Wo	E
Ektodermale Dysplasie (ECTD) * ECTD1, ECTD2, ECTD3, ECTD9, ECTD10A, ECTD10B	P, M	305100 129500 189500 614931 129490 224900	EDA, GJB6, MSX1, HOXC13, EDAR	2-4 Wo	E
Ehlers-Danlos-Syndrom, Typ IV (EDS4) S	P, M	130050	COL3A1	2-4 Wo	E
Emery-Dreifuss-Muskeldystrophie (EDMD)* EDMD1, EDMD2, EDMD3, EDMD6, EDMD7	P, M	310300 181350 300396 614302	EMD, LMNA, FHL1, TMEM43	2-4 Wo	E
Epidermolysis bullosa (EB) EB junctionalis (Non-Herlitz-Typ, Herlitz-Typ), EB simplex (lokalisiert, generalisiert, Dowling-Meara-Typ), EB akantholytisch letal	P, M	226650 226700 131800 131900 131760 600100 609638	LAMB3, KRT14, DSP	2-4 Wo	E
Epileptische Enzephalopathie, frühinfantile (EIEE) EIEE1, EIEE2, EIEE3, EIEE4, EIEE5, EIEE6, EIEE7, EIEE9, EIEE10, EIEE11, EIEE14, EIEE16, EIEE19, EIEE27	P, M	308350 300672 609304 612164 613477 607208 613720 300088 613402 613721 614959 615338 615744 616139	ARX, CDKL5, SLC25A22, STXBP1, SPTAN1, SCN1A, KCNQ2, PCDH19, PNKP, SCN2A, KCNT1, TBC1D24, GABRA1, GRIN2B	2-5 Wo	E
Epilepsie, Pyridoxin-abhängige (EPD) *	P	266100	ALDH7A1	2-3 Wo	E
Epiphysäre Dysplasie, multiple, Typ 4 (EDM4)	P	226900	SLC26A2	2-3 Wo	E
Episodische Ataxie (EA) * EA1, EA2, EA5, EA6	P, M	160120 108500 613855 612656	KCNA1, CACNA1A, CACNB4, SLC1A3	2-4 Wo	E
Epstein-Syndrom (EPSTNS)	P	153650	MYH9	2-4 Wo	E
Erythermalgie, primäre	P	133020	SCN9A	2-3 Wo	E
Erythrozytose 2, familiäre (ECYT2)	P, M	263400	VHL	2-3 Wo	E
Exostosen, multiple (EXT)	P, M	133700 133701	EXT1, EXT2	2-3Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
F					
Fabry-Krankheit	P, M	301500	GLA	2-3 Wo	E
Fallot-Tetralogie (TOF)	P, M	187500	JAG1, TBX1, GATA4, NKX2-5, ZFPM2	3-4 Wo	E
Familiäre adenomatöse Polyposis (FAP) * FAP1, FAP2, FAP3	P, M	175100 608456 616415	APC, MUTYH, NTHL1	2-4 Wo	E
Familiäres kälteinduziertes autoinflammatorisches Syndrom 1 (FCAS1)	P	120100	NLRP3	2-3 Wo	E
Familiäres Mittelmeerfieber (FMF) *	P, M	249100 134610	MEFV	3-5 Wo	E
Fanconi-Anämie (FA) * FANCC, FANCD1, FANCD2, FANCI, FANCJ, FANCN, FANCO, FANCS	P, M	227645 605724 227646 609053 609054 610832 613390 617883	FANCC, BRCA2, FANCD2, FANCI, BRIP1, PALB2, RAD51C, BRCA1	2-4 Wo	E, FW, C
Fechtner-Syndrom (FTNS)	P	153640	MYH9	2-4 Wo	E
Feingold-Syndrom	P	164280	MYCN	2-3 Wo	E
FG-Syndrom FGS1, FGS2, FGS4	P	305450 300321 300422	MED12, FLNA, CASK	2-5 Wo	E
Fieberkrämpfe, familiäre (FEB) FEB3A, FEB3B, FEB4, FEB8, FEB11	P, M	604403 613863 604352 611277 609562	SCN1A, SCN9A, ADGRV1 (GPR98), GABRG2, CPA6	2-5 Wo	E
FILS-Syndrom	P	615139	POLE	2-3 Wo	E
Fischaugen-Krankheit (FED)	P	136120	LCAT	2-3 Wo	E
Floating-Harbor-Syndrom (FLHS) S	P	136140	SRCAP	4-6 Wo	E
Fokal segmentale Glomerulosklerose (FSGS) FSGS1, FSGS2, FSGS5	P	603278 603965 613237	ACTN4, TRPC6, INF2	2-4 Wo	E
Fokale Epilepsie mit variablen Herden (FFEVF) FFEVF1, FFEVF3	P	604364 617118	DEPDC5, NPRL3	2-4 Wo	E
Fragiles X-Syndrom	F, B	300624	FMR1	2-3 Wo	E, FW, C
Fragiles X-Tremor/Ataxie-Syndrom /FXTAS) CO	F, B	300623	FMR1	2-3 Wo	E
Frasier-Syndrom	P, M	136680	WT1	2-3 Wo	E
Friedreich-Ataxie 1 (FRDA1) CO	F	606829	FRDA	2-3 Wo	E, FW, C
Frontonasale Dysplasie 1 (FND1)	P	136760	ALX3	2-3 Wo	E
Fruktose-1,6-Bisphosphatase-Mangel (FBP1D)	P, M	229700	FBP1	2-3 Wo	E
Fruktoseintoleranz, hereditäre (HFI) *	P, M	229600	ALDOB	2-3 Wo	E
Fumarase-Mangel	P, M	606812	FH	2-3 Wo	E
G					
Galaktosämie Galaktosämie I, Galaktosämie II, Galaktosämie III	P, M	230400 230200 230350	GALT, GALK1, GALE	2-3 Wo	E
Gallenblasenerkrankung (GBD) GBD1, GBD4	P; M	600803 611465	ABCB4, ABCG8	2-4 Wo	E
Gardner-Syndrom (GS) *	P	175100	APC	2-3 Wo	E
Gastrointestinaler Stromatumor (GIST) *	P, M	606764	KIT, SDHB, SDHD	2-4 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Gaucher-Krankheit (GD)	P, M	230800 230900 231000 231005 608013	GBA	2-3 Wo	E
Geleophysische Dysplasie 2 (GPHYSD2) *	P, M	614185	FBN1	3-6 Wo	E
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFSP) GEFSP1, GEFSP2, GEFSP3, GEFSP5, GEFSP7, GEFSP9	P, M	604233 604403 611277 613060 613863 616172	SCN1B, SCN1A, GABRG2, GABRD, SCN9A, STX1B	2-4 Wo	E
Generalisierte idiopathische Epilepsie (EIG) * EIG6 (ECA6), EIG8, EIG9 (EJM6), EIG10 (EJM7), EIG11 (EJM8, EJA2), EIG12, EIG13 (EJM5, ECA4), ECA2	P, M	611942 612899 607682 613060 607628 614847 611136 607681	CACNA1H, CASR, CACNB4, GABRD, CLCN2, SLC2A1, GABRA1, GABRG2	2-4 Wo	E
Gilbert-Syndrom *, S	P, F	143500	UGT1A1	2-3 Wo	E
Glanzmann-Thrombasthenie (GT)	P	273800	ITGA2B, ITGB3	3-4 Wo	E
Gliedergürtelmuskeldystrophie (LGMD, MDDGC) * LGMDR4 (LGMD2E), LGMDR5 (LGMD2C), LGMDR10 (LGMD2J), LGMDR9 (MDDGC5, LGMD2I), LGMDR13 (MDDGC4, LGMD2M), LGMDR20 (MDDGC7, LGMD2U)	P, M	604286 253700 608807 611588 607155 616052	SBCB, SGCG, TTN, FKRP, FKTN, ISPD	2-6 Wo	E
GLUT1-Mangel-Syndrom (GLUT1DS) * GLUT1DS1, GLUT1DS2	P, M	606777 612126	SLC2A1	2-3 Wo	E
Glycerol-Kinase-Mangel (GKD)	P	307030	GK	2-3 Wo	E
Glycin-Enzephalopathie (GCE) *	P, M	605899	AMT, GLDC, GCSH	2-4 Wo	E
Glykogenspeicherkrankheit des Herzens *	P	261740	PRKAG2	2-3 Wo	E
Glykogenspeicherkrankheit (GSD)	P, M	232300 232700	GAA, PYGL	2-3 Wo	E
Granulomatose, chronische, X-chromosomal (CGDX) *	P, M	306400	CYBB	2-4 Wo	E
Gray-Platelet-Syndrom (GPS)	P	139090	NBEAL2	4-6 Wo	E
Greig-Zephalopolysyndaktylie-Syndrom (GCPS)	P, M	175700	GLI3	2-4 Wo	E
H					
Hämoglobin-F-Persistenz, hereditäre (HPFH) *	P, M	141749	HBB	2-3 Wo	E
Hajdu-Cheney-Syndrom (HJCYS)	P	102500	NOTCH2	2-3 Wo	E
Hämaturie, benigne familiäre (BFH) *	P, M	141200	COL4A3, COL4A4	2-5 Wo	E
Hämochromatose (HFE) S HFE1, HFE2A, HFE2B, HFE3, HFE4, HFE5	P, M	235200 602390 613313 604250 606069	HFE, HJV, HAMP, TFR2, SLC40A1, FTH1	2-4 Wo	E
Hämolytisch-urämisches Syndrom, atypisches (AHUS) AHUS3, AHUS7	P	612923 615008	CFI, DGKE	2-4 Wo	E
HARP-Syndrom	P, M	607236	PANK2	3-4 Wo	E
Heinz-Körper-Anämie *	P, M	140700	HBB	2-3 Wo	E
Hepatische Lipase-Mangel (HL-Mangel)	P	614025	LIPC	2-3 Wo	E
Hereditäre motorisch-sensible Neuropathie, Typ 6A (HMSN6A)	P, M	601152	MFN2	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Hereditäres nicht-polypöses kolorektales Karzinom (HNPCC) *, S, CO HNPCC1, HNPCC2, HNPCC3, HNPCC4, HNPCC5, HNPCC6, HNPCC7, HNPCC8	P, M, MSI, I Material beachten (T)	120435 609310 614337 614350 614331 614385 613244	MSH2, MLH1, PMS1, PMS2, MSH6, TGFBR2, MLH3, EPCAM	2-3 Wo	E T: in Paraffin eingebettete Tumorblöcke des Patienten (Primärtumor)
Hirntumor-Polyposis-Syndrom (BTPS) * BTPS1, BTPS2	P, M	276300 175100	MLH1, MSH2, MSH6, PMS2, APC	2-4 Wo	E
Hirschsprung-Krankheit, Typ 1 (HSCR1)	P, M	142623	RET	2-3 Wo	E
Holocarboxylase-Synthetase-Mangel	P	253270	HLCS	2-4 Wo	E
Holoprosenzephalie (HPE) HPE3, HPE7	P, M	142945 610828	SHH, PTCH1	2-3 Wo	E
Holt-Oram-Syndrom (HOS)	P	142900	TBX5	2-3 Wo	E
Homocystinurie durch CBS-Mangel	P	236200	CBS	2-4 Wo	E
Homocystinurie durch MTHFR-Aktivitätsmangel *, CO	P	236250	MTHFR, 677C>T	2 Wo	E
Huntington-Krankheit (HD) *, 1	F	143100	HTT	2 Wo	E, FW, C
Hutchinson-Gilford-Progerie-Syndrom (HGPS) *	P, M	176760	LMNA	2-3 Wo	E
Hydrozephalus mit Aquäduktstenose (HSAS)	P, M	307000	L1CAM	2-4 Wo	E
Hyperalphalipoproteinämie (HALP) HALP1, HALP2	P	143470 614028	CETP, APOC3	2-3 Wo	E
Hyperbilirubinämie (HBLR)	P	237900 143500 606785 218800	UGTA1	2-3 Wo	E
Hypercholesterinämie, familiäre (FHCL) * FHCL1, FHCL2, FHCL3, FHCL4	P, M	143890 144010 603776 603813	LDLR, APOB (Apo-B-100-Allel), PCSK9, LDLRAP1	2-4 Wo	E
Hyperchylomikronämie *	P, M	238600 207750 615947 144650	LPL, GPIHBP1, APOC2, APOA5	2-3 Wo	E
Hyperekplexie (HKPX) HKPX1, HKPX4	P	149400 618011	GLRA1, ATAD1	2-4 Wo	E
Hyperinsulinämische Hypoglykämie, familiäre (HHF) *, S HHF1, HHF2, HHF3	P	256450 601820	ABCC8, KCNJ11, GCK	2-4 Wo	E
Hyper-IgD-Syndrom (HIDS)	P	260920	MVK	2-3 Wo	E
Hyperkaliämische periodische Paralyse (HYPP)	P	170500	SCN4A	2-3 Wo	E
Hyperkalzämie, infantile (HCINF) HCINF1, HCINF2	P	143880 616963	CYP24A1, SLC34A1	2-3 Wo	E
Hyperlipoproteinämie Typ III *, S	P	107741	APOE	2-3 Wo	E
Hyperlipoproteinämie (HLP) * Typ IA, Typ IB, Typ ID, Typ IIA, Typ III, Typ V	P, M	238600 207750 615947 143890 617347 144650	LPL, APOC2, GPIHBP1, LDLR, APOE, APOA5	2-4 Wo	E
Hypertriglyzeridämie *	P, M	238600 615947 207750 144650 145750 246650	LPL, GPIHBP1, APOC2, APOA5, LMF1	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Hypertrophe Dejerine-Sottas-Neuropathie (DSS) *	P, M	145900	PMP22, MPZ, EGR2, GJB1	2-4 Wo	E
Hypertrophe Kardiomyopathie (HCM, CMH) * CMH1, CMH2, CMH3, CMH4, CMH5, CMH6, CMH7, CMH8, CMH9, CMH10, CMH11, CMH12, CMH13, CMH14, CMH15, CMH17, CMH20, CMH22, CMH23, CMH	P, M	192600 115195 115196 115197 600858 613690 608751 613765 608758 612098 612124 613243 613251 613255 613873 613876 615248 612158	MYH7, TNNT2, TPM1, MYBPC3, PRKAG2, TNNI3, MYL3, TTN, MYL2, ACTC1, CSRP3, TNNC1, MYH6, VCL, JPH2, NEXN, MYPN, ACTN2, CAV3	4-6 Wo	E
Hypoalphalipoproteinämie, familiäre (FHA)	P	604091	ABCA1, APOA1	2-3 Wo	E
Hypobetalipoproteinämie, familiäre (FHBL) *	P	615558	APOB	2-3 Wo	E
Hypochondroplasie (HCH) *, S	P	146000	FGFR3	2-3 Wo	E, FW, C
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (HH) HH1, HH2, HH5, HH8, HH10, HH12	P, M	308700 147950 612370 614837 614839 614841	KAL1, FGFR1, CHD7, KISS1R, TAC3, GNRH1	2-4 Wo	E
Hypokaliämische periodische Paralyse, Typ 2 (HOKPP2)	P	613345	SCN4A	2-3 Wo	E
Hypokalziurische Hyperkalzämie (HHC) HHC1, HHC2, HHC3	P	145980 145981 600740	CASR, GNA11, AP2S1	2-4 Wo	E
Hypoplastisches Linksherz-Syndrom (HLHS) HLHS1, HLHS2	P, M	241550 614435	GJA1, NKX2-5	2-3 Wo	E
Hypospadie 1, X-chromosomal (HYSP1)	P, M	300633	AR	2-4 Wo	E
I					
Infantile Myofibromatose (IMF) IMF1, IMF2	P, M	228550 615293	PDGFRB, NOTCH3	2-4 Wo	E
Iridogoniodysgenesie (IRID) IRID1, IRID2	P, M	601631 137600	FOXC1, PITX2	2-4 Wo	E
Isolierter Wachstumshormonmangel (IGHD) IGHD1A, IGHD1B, IGHD2, IGHD3, IGHD5	P, M	262400 612781 173100 307200 618160	GH1, BTK, RNPC3	2-4 Wo	E
Isovalerianazidämie (IVA) *	P	243500	IVD	2-3 Wo	E
J					
Jackson-Weiss-Snydrom (JWS) *, S	P, M	123150	FGFR2	2-4 Wo	E
Jervell- und Lange-Nielsen-Syndrom (JLNS) * JLNS1, JLNS2	P, M	220400 612347	KCNQ1, KCNE1	2-4 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Joubert-Syndrom (JBTS) JBTS1, JBTS2, JBTS4, JBTS5, JBTS6, JBTS7, JBTS9, JBTS10, JBTS13, JBTS27, JBTS28, JBTS32	P, M	213300 608091 609583 610188 610688 611560 612285 300804 614176 617120 617121 617757	INPP5E, TMEM216, NPHP1, CEP290, TMRM67, RPGRIP1L, CC2D2A, OFD1, TCTN1, B9D1, MKS1; SUFU	2-5 Wo	E, FW, C
Juvenile myoklonische Epilepsie (EJM) EJM5, EJM6, EJM7, EJM8	P	611136 607682 613060 607628	GABRA1, CACNB4, GABRD, CLCN2	2-4 Wo	E
Juveniles Polyposis-Syndrom (JPS)	P, M	174900	BMPR1A, SMAD4 (MADH4)	2-3 Wo	E
K					
Kabuki-Syndrom (KABUK) KABUK1, KABUK2	P, M	147920 300867	KMT2D (MLL2), KDM6A	3-4 Wo	E
Kallmann-Syndrom KAL1, KAL2	P, M	308700 147950	KAL1, FGFR1	2-4 W	E
Kampomele Dysplasie (CMPD)	P, M	114290	SOX9	2-3 Wo	E
Kardiofaziokutanes Syndrom (CFC) * CFC1, CFC2, CFC3, CFC4	P, M	115150 615278 615279 615280	BRAF, KRAS, MAP2K1, MAP2K2	2-4 Wo	E, FW, C
Kartagener-Syndrom * CILD1, CILD3, CILD7, CILD	P, M	244400 608644 611884	DNAI1, DNAH5, DNAH11, DNAH8	3-6 Wo	E, FW, C
Katarakt (CTRCT) CTRCT34, CTCTR40	P	612968 302200	FOXE3, NHS	2-4 Wo	E
Keratitis, hereditäre	P, M	149190	PAX6	2-3 Wo	E
Keratitis-Ichthyosis-Taubheit-Syndrom (KID-Syndrom) *	P	148210	GJB2	2-3 Wo	E
Keratokonus 1 (KTCN1)	P	148300	VSX1	2-3 Wo	E
Kleidokraniale Dysplasie (CCD)	P, M	119600	RUNX2	2-3 Wo	E
Kleinwuchs, idiopathischer, X-chromosomal (ISS) *	P, M	300582	SHOX, SHOXY	2-3 Wo	E
Klippel-Feil-Syndrom (KFS) KFS1, KFS2	P	118100 214300	GDF6, MEOX1	2-3 Wo	E
Kohlschutter-Tonz-Syndrom (KTZS)	P	226750	ROGDI	2-3 Wo	E
Kolobom des Sehnerven	P, M	120430	PAX6	2-3 Wo	E
Kongenitale Anomalien der Niere und des Harntraktes (CAKUT) CAKUT1, CAKUT2	P	610805 143400	DSTYK, TBX18	2-4 Wo	E
Kongenitale bilaterale Aplasie des Vas deferens (CBAVD) *, S	N, M	277180	CFTR	2-4 Wo	E
Kongenitale Glykosylierungsstörung (CDG) CDG1K, CDG2G	P	608540 611209	ALG1, COG1	2-4 Wo	E
Kongenitaler Herzfehler, multipler (CHTD) CHTD4, CHTD5, CHTD6	P	615779 617912 613854	NR2F2, GATA5, GDF1	2-4 Wo	E
Koronare Herzkrankheit (CAD), Prädisposition *	P	152200	LPA (rs10455872; rs3798220)	1-2 Wo	E
Kortikale Dysplasie, komplexe, mit anderen Hirnfehlbildungen	P	614039	TUBB3	2-3 Wo	E
Krabbe-Krankheit	P, M	245200	GALC	2-4 Wo	E, FW, C
Kraniosynostose (CRS) CRS1, CRS3, CRS4, CRS7	P, M	123100 615314 600775 617439	TWIST, TCF12, ERF, SMAD6	2-4 Wo	E, FW, C

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
L					
LADD-Syndrom *, S	P, M	149730	FGFR2, FGFR3	2-3 Wo	E
Laktase-Mangel, kongenitaler	P	223000	LCT	2-4 Wo	E
Laktose-Intoleranz, adulter Typ *, CO	P, S	223100	MCM6	2 Wo	E
Langer mesomele Dysplasie (LMD) *	P, M	249700	SHOX, SHOXY	2-3 Wo	E
Laron-Syndrom	P, M	262500	GHR	2-3 Wo	E
Lebersche kongenitale Amaurose (LCA) LCA4, LCA10, LCA13, LCA17, LCA18	P	604393 611755 612712 615360 608133	AIPL1, CEP290, RDH12, GDF6, PRPH2	2-5 Wo	E
Lecithin-Cholesterin-Acyltransferase-Mangel (LCAT-Mangel)	P	245900	LCAT	2-3 Wo	E
Legius-Syndrom	P, M	611431	SPRED1	2-3 Wo	E
Leiomyomatose und Nierenzellkarzinom, hereditäre (HLRCC)	P, M	150800	FH	2-3 Wo	E
LEOPARD-Syndrom * LPRD1, LPRD2, LPRD3	P	151100 611554 613707	PTPN11, RAF1, BRAF	2-4 Wo	E
Leri-Weill Dyschondrosteose (LWD) *	P, M	127300	SHOX, SHOXY	3-4 Wo	E
Leukodystrophie, hypomyelinisierende (HLD) HLD1, HLD2, HLD3, HLD9, HLD11, HLD12, HLD14	P, M	312080 608804 260600 616140 616494 616683 617899	PLP1, GJC2 (GJA12), AIMP1, RARS, POLR1C, VPS11, UFM1	2-4 Wo	E, FW, C
Leukoenzephalopathie mit Dystonie und motorischer Neuropathie	P	613724	SCP2	2-3 Wo	E
Lhermitte-Duclos-Syndrom (LDD) *	P, M	158350	PTEN	2-3 Wo	E
Li-Fraumeni-Syndrom (LFS) LFS1, LFS2	P, M	151623 609265	TP53,CHEK2	2-4 Wo	E, FW, C
Linksventrikuläre Noncompaction-Kardiomyopathie (LVNC) LVNC4, LVNC5, LVNC6, LVNC8, LVNC9, LVNC10	P, M	613424 613426 601494 615373 611878 615396	ACTC1, MYH7, TNNT2, TPM1, MYBPC3	2-4 Wo	E
Lipasemangel, kombinierter *	P	146650	LMF1	2-3 Wo	E
Lipodystrophie, familiäre partielle (FPLD) * FPLD2, FPLD6	P, M	151660 615980	LMNA, LIPE	2-3 Wo	E
Lipodystrophie, kongenitale generalisierte (CGL) CGL1, CGL2	P	608594 269700	AGPAT2, BSCL2	2-3 Wo	E
Lipoprotein(a)-Erhöhung *	P	152200	LPA (rs10455872; rs3798220)	2-3 Wo	E
Lipoprotein-Glomerulopathie (LPG) *	P	611771	APOE	2-3 Wo	E
Lissenzephalie 1 (LIS1)	P, M	607432	PAFAH1B1	2-4 Wo	E, FW, C
Lissenzephalie, X-chromosomal (LISX) LISX1, LISX2	P, M	300067 300215	DCX, ARX	2-4 Wo	E, FW, C
Long-QT-Syndrom (LQT) * LQT1, LQT2, LQT3, LQT5, LQT6, LQT7, LQT8, LQT9	P, M	192500 613688 603830 613695 613693 170390 601005 618447 611818	KCNQ1, KCNH2,SCN5A, KCNE1, KCNE2, KCNJ2, CACNA1C, CAV3	2-5 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Loeys-Dietz-Syndrom (LDS) * LDS1, LDS2, LDS3, LDS4, LDS5 *Hinweis: Bei Kassenpatienten erfolgt eine Diagnostik gemäß EBM Kapitel 11.4.2.*	P, M	609192 610168 613795 614816 615582	TGFBR1, TGFBR2, SMAD3 TGFB2, TGFB3	2-4 Wo	E
Lowe okulo-zerebro-renales Syndrom (OCRL)	P	309000	OCRL	2-3 Wo	E
Lujan-Fryns-Syndrom	P	309520	MED12	2-3 Wo	E
Lymphödem, hereditäres (LMPH1) LMPH1A, LMPH1C, LMPH1D	P	153100 613480 615907	FLT4, GJC2, VEGFC	2-4 Wo	E
Lymphoproliferatives Syndrom 1, X-chromosomales (XLP1)	P, M	308240	SH2D1A	2-3 Wo	E
Lynch-Syndrom * Falls Tumormaterial vorliegt, findet zuvor eine Untersuchung der Mikrosatelliteninstabilität bzw. eine immunhistochemische Untersuchung statt. CO *Hinweis: Mikrosatelliteninstabilität und immunhistochemische Untersuchungen an Tumorgewebe sind keine akkreditierten Verfahren.*	P, M, MSI, I Material beachten (T)	120435	MSH2, MLH1, PMS2, MSH6	2-3 Wo	E T: in Paraffin eingebettete Tumorblöcke des Patienten (Primärtumors)
Lysosomale saure Lipase-Mangel (LAL-Mangel)	P	278000	LIPA	2-3 Wo	E
M					
Magenkarzinom, hereditäres diffuses (HDGC) *	P, M	137215	CDH1	2-3 Wo	E
Makuladegeneration, altersbedingte (ARMD) ARMD2, ARMD7, ARMD13	P, M	153800 610149 615439	ABCA4, HTRA1, CFI	2-4 Wo	E
Maligne Hyperthermie 1 (MHS1) S	P	145600	RYR1	3-6 Wo	E
Malouf-Syndrom *	P, M	612112	LMNA	2-3 Wo	E
Marfan-Syndrom (MFS) *, S	P, M	154700	FBN1	2-4 Wo	E
Marshall-Syndrom (MRSHS)	P, M	154780	COL11A1	3-4 Wo	E
MASA-Syndrom	P, M	303350	L1CAM	2-4 Wo	E
MASS-Syndrom *	P, M	604308	FBN1	3-6 Wo	E
Mastozytose	P, M	154800	KIT	2-3 Wo	E
McKusick-Kaufman-Syndrom (MKKS)	P	236700	MKKS	2-3 Wo	E
May-Hegglin-Anomalie (MHA)	P	155100	MYH9	2-4 Wo	E
Meckel-Syndrom (MKS) MKS1, MKS2, MKS3, MKS4, MKS5, MKS6, MKS9	P	249000 603194 307361 611134 611561 612284 614209	MKS1, TMEM216, TMEM67, CEP290, RPGRIP1L, CC2D2A, B9D1	2-4 Wo	E, FW, C
Megalenzephalie-Polymikrogyrie-Polydaktylie-Hydrocephalus-Syndrom 2 (MPPH2)	P	615937	AKT3	2-3 Wo	W
Melanom-Pankreaskarzinom-Syndrom	P, M	606719	CDKN2A	2-3 Wo	E
Menkes-Krankheit (MK, MNK)	P, M	309400	ATP7A	2-3 Wo	E
Mentale Retardierung mit Sprachstörung	P, M	613670	FOXP1	2-3 Wo	E
Mentale Retardierung und Mikrozephalie mit pontozerebellärer Hypoplasie (MICPCH)	P	300749	CASK	2-4 Wo	E
Mentale Retardierung, autosomal-rezessiv, 5 (MRT5)	P	611091	NSUN2	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Mentale Retardierung, X-chromosomal (MRX) MRXARX, MRX19, MRX21, MRX63, MRX94, MRX97	P, M	300419 300844 300143 300387 300699 300803	ARX, RPS6KA3, IL1RAPL1, ACSL4, GRIA3 ZNF711	2-4 Wo	E
Mentale Retardierung, autosomal-dominant (MRD) MRD1, MRD5, MRD6, MRD7, MRD12, MRD15, MRD19, MRD20, MRD22, MRD29	P, M	156200 612621 613970 614104 135900 614608 615075 613443 616078	MBD5, SYNGAP1, GRIN2B, DYRK1A, ARID1B, SMARCB1, CTNNB1, MEF2C, ZBTB18, SETBP1	2-4 Wo	E
Mevalon-Acidurie (MEVA)	P	251170	MVK	2-3 Wo	E
Migräne, familiäre hemiplegische (FHM) *, S FHM1, FHM2, FHM3	P, M	141500	ATP1A2, CACNA1A, SCN1A	2-5 Wo	E
Mikrozephalie, primäre, autosomal-rezessiv (MCPH) MCPH1, MCPH2, MCPH3, MCPH4, MCPH5, MCPH6, MCPH7, MCPH9	P, M	251200 604317 604804 604321 608716 608393 612703 614852	MCPH1, WDR62, CDK5RAP2, CASC5,ASPM, CENPJ, STIL, CEP152	4-6 Wo	E
Miller-Dieker-Lissenzephalie-Syndrom (MDLS)	M	247200 613215	17p13.3-MDCR	2-3 Wo	E, FW, C
Minicore-Myopathie mit externer Ophtalmoplegie	P	255320	RYR1	4-6 Wo	E
Mitochondriales DNA-Depletionssyndrom 4 (MTDPS4) MTDPS4A, MTDPS4B	P, M	203700 613662	POLG	2-4 Wo	E
MLCRD-Syndrom	P	152950	KIF11	3-4 Wo	E
MMR-Defizienz-Syndrom (MMRCS) * MMRCS1, MMRCS2, MMRCS3,MMRCS4	P, M	276300 619096 619097 619101	MLH1, MSH2, MSH6, PMS2	2-4 Wo	E
MODY (Maturity-Onset Diabetes of the Young) CO MODY1, MODY2, MODY3, MODY4, MODY5, MODY10, MODY13	P, M	606391 125850 125851 606392 600496 137920 613370	HNF4A, GCK, HNF1A, PDX1, HNF1B, INS, KCNJ11	2-3 Wo	E
Mohr-Tranebjaerg-Syndrom (MTS)	P	304700	TIMM8A	2-4 Wo	E
Morbus Meulengracht	P	143500	UGT1A1	2-3 Wo	E
Mowat-Wilson-Syndrom	P, M	235730	ZEB2	2-3 Wo	E
Muckle-Wells-Syndrom (MWS)	P	191900	NLRP3	2-3 Wo	E
Muenke-Syndrom (MNKES) *	P	602849	FGFR3	2-3 Wo	E
Muir-Torre-Syndrom (MRTES) *	P, M	158320	MLH1, MSH2	2-3 Wo	E
Müller-Gang-Persistenzsyndrom (PMDS) * Typ 1, Typ 2	P	261550	AMH, AMHR2	2-4 Wo	E
Multiple endokrine Neoplasie (MEN) MENI, MENIIA, MENIIB	P, M	131100 162300 171400	MEN1, RET	2-4 Wo	E, FW, C
Multiples Pterygium-Syndrom, Escobar-Variante (EVMPS)	P	265000	CHRNG	2-3 Wo	E
Multiples Pterygium-Syndrom, letaler Typ (LMPS)	P	253290	CHRNG, CHRND, CHRNA1	2-4 Wo	E
Muskeldystrophie, Typ Becker (BMD) S	P, M	300376	DMD	2-4 Wo	E, FW, C
Muskeldystrophie, Typ Duchenne (DMD) S	N, M, P	310200	DMD	2-4 Wo	E, FW, C

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Myasthenes Syndrom, kongenitales (CMS) CMS1A, CMS1B, CMS2A, CMS2B, CMS2C, CMS3A, CMS3B, CMS3C, CMS11, CMS16	P	601462 608930 616313 616314 616321 616322 616323 616326 614198	CHRNA1, CHRNB1, CHRND, RAPSN, SCN4A	2-4 Wo	E
Myeloperoxidase-Mangel (MPOD)	P	254600	MPO	2-3 Wo	E
Myotone Dystrophie 1 (DM1) [CO]	F	160900	DMPK	2-3 Wo	E, FW, C
Myotonia congenita, Typ Becker, autosonal-rezessiv	P	255700	CLCN1	2-3 Wo	E
Myotonia congenita, Typ Thomsen, autosomal-dominant	P	160800	CLCN1	2-3 Wo	E
N					
Nebennierenrinden-Karzinom, hereditäres (ADCC)	P, M	202300	TP53	2-3 Wo	E
Nephronophthise (NPHP) NPHP1, NPHP11	P, M	256100 613550	NPHP1, TMEM67	2-4 Wo	E, FW, C
Nephrotisches Syndrom (NPHS) NPHS2, NPHS4, NPHS6, NPHS7	P, M	600995 256370 614196 615008	NPHS2, WT1, PTPRO, DGKE	2-4 Wo	E
Neugeborenenkrämpfe, benigne familiäre (BFNS) BFNS1, BFNS2	P, M	121200 121201	KCNQ2, KCNQ3	2-3 Wo	E
Neurodegeneration mit Eisenakkumulation im Gehirn 1 (NBIA1)	P, M	607236	PANK2	3-4 Wo	E
Neurofibromatose, Typ 1 (NF1) NF1	P, M	162200	NF1	2-4 Wo	E, FW, C
Neurofibromatose-Noonan-Syndrom (NFNS) [*]	P, M	601321	NF1	3-4 Wo	E
Neuropathie, hereditäre, mit Neigung zu Drucklähmungen [*, S] (HNPP)	P, M	162500	PMP22	2-4 Wo	E
Neuropathie, hereditäre sensorische und autonome (HSAN) HSAN1A, HSAN2D, HSAN3	P	162400 143000 223900	SPTLC1, SCN9A, IKBKAP	2-4 Wo	E
Neuropathie, kongenitale hypomyelinisierende (CHN) [*] CHN1, CHN2	P, M	605253 618184	EGR2; MPZ	2-4 Wo	E
Neutropenie, schwere kongenitale (SCN) SCN1, SCN3, SCNX	P	202700 610738 300299	ELANE, HAX1, WAS	2-4 Wo	E
Nicolaides-Baraitser-Syndrom (NCBRS)	P	601358	SMARCA2	2-3 Wo	E
Nierenzellkarzinom 1, papilläres (RCCP1)	P	605074	MET	2-3 Wo	E
Nijmegen-Breakage-Syndrom	P	251260	NBN (NBS1)	2-3 Wo	E
Nystagmus, kongenitaler (NYS) NYS1, NYS6	P	310700 300814	FRMD7, GPR143	2-4 Wo	E
Noonan-ähnliches Syndrom mit losem Anagenhaar (NSLH)	P	607721	SHOC2	2-3 Wo	E
Noonan-Syndrom (NS) [*, S] NS1, NS2, NS3, NS4, NS5, NS6, NS7, NS8, NS9, NS10, NS11 Es erfolgt eine Stufendiagnostik gemäß EBM Kapitel 11.4.2.	P, M	163950 605275 609942 610733 611553 613224 613706 615355 616559 616564 618499	PTPN11, LZTR1KRAS, SOS1, RAF1, NRAS, BRAF, SOS2, RIT1, MRAS	2-4 Wo	E, FW, C

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
O					
Odonto-onycho-dermale Dysplasie (OODD)	P	257980	WNT10A	2-3 Wo	E
Okulodentodigitale Dysplasie (ODDD)	P, M	164200 257850	GJA1	2-3 Wo	E
Okzipitalhorn-Syndrom (OHS)	P, M	309400	ATP7A	2-3 Wo	E
Opitz-GBBB-Syndrom, Typ 1 (GBBB1)	P, M	300000	MID1	2-3 Wo	E
Opitz-Kaveggia-Syndrom (OKS)	P	305450	MED12	2-3 Wo	E
Optikusatrophie (OPA) OPA1, OPA3	P, M	165500 165300	OPA1, OPA3	2-3Wo	E
Orofaziale Spalte OFC5, OFC6	P, M	608874	MSX1, IRF6	2-3 Wo	E, FW, C
Osteogenesis imperfecta (OI) * OI Typ I, II, III, IV, V, VII, VIII, XV	P, M	166200 166210 259420 166220 610967 610682 610915 615220	COL1A1, COL1A2, IFITM5, CRTAP, LEPRE1, WNT1	2-5 Wo	E, FW, C
Osteogenesis imperfecta und Ehlers-Danlos-Syndrom, kombiniert (OIEDS) *	P, M	619115 619120	COL1A1, COL1A2	2-4 Wo	E
Ovarialdysgenesie (ODG) ODG1, ODG2	P	233300 300510	FSHR, BMP15	2-3 Wo	E
Ovarielles Hyperstimulationssyndrom (OHSS)	P	608115	FSHR	2-3 Wo	E
P					
Palmoplantarkeratose mit Hörverlust *	P, M	148350	GJB2	2-3 Wo	E
Panhypopituitarismus, X-chromosomal (PHPX)	P	312000	SOX3	2-3 Wo	E
Pankreas-Agenesie 1 (PAGEN1)	P, M	260370	PDX1	2-3 Wo	E
Pankreatitis, hereditäre (PCTT) * PCTT, Idiopatische PCTT, Prädisposition für PCTT	P, M	167800	PRSS1, SPINK1, CFTR, CTRC	2-4 Wo	E
Paragangliom (PGL) * PGL1, PGL4, PGL5	P, M	168000 115310 614165	SDHD, SDHB, SDHA	2-4 Wo	E
Parkinson-Krankheit (PARK) PARK1, PARK2, PARK4, PARK8	P, M	168601 600116 605543 607060	SNCA, PARK2 (PRKN), LRRK2	2-3 Wo	E
Partington-Syndrom (PRTS)	P, M	309510	ARX	2-3 Wo	E
PCWH-Syndrom (PCWH)	P, M	609136	SOX10	2-3 Wo	E
Pelizaeus-Merzbacher-Krankheit (PMD)	P, M	312080	PLP1	2-3 Wo	E, FW
Periodische Fiebersyndrome * FMF, FPF, TRAPS, FCAS1 (CAPS1), MWS (CAPS2), CINCA (CAPS3), HIDS, MEVA	P, M	249100 134610 142680 120100 191900 607115 249100 260920	MEFV, TNFRSF1A, NLRP3, MVK	2-4 Wo	E
Peroxisomale Biogenesestörung 9B (PBD9B)	P	614879	PEX7	2-4 Wo	E
Peutz-Jeghers-Syndrom (PJS)	P, M	175200	STK11	2-3 Wo	E
Pfeiffer-Syndrom *, S	P, M	101600	FGFR1, FGFR2	2-3 Wo	E, FW, C
Phäochromozytom *	P, M	171300	SDHD, SDHB, RET, VHL, SDHA	2-4 Wo	E
Piebaldismus	P, M	172800	KIT, SNAI2	2-3 Wo	E
Pitt-Hopkins-Syndrom (PTHS)	P, M	610954	TCF4	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Pitt-Hopkins-ähnliches Syndrom (PTHSL) PTHSL1, PTHSL2	P, M	610042 614325	CNTNAP2, NRXN1	2-4 Wo	E
Pneumothorax, primärer spontaner (PSP)	P	173600	FLCN	2-3 Wo	E
Polymikrogyrie, symmetrische oder asymmetrische (PMGYSA)	P	610031	TUBB2B	2-3 Wo	E
Polyzystische Nieren-Erkrankung (PKD) PKD1, PKD2, PKD4, PKD5, PKD6	P, M	173900 613095 600666 263200 617610 618061	PKD1, PKD2, GANAB, PKHD1, DZIP1L, DNAJB11	2-5 Wo	E
Pompe-Krankheit	P	232300	GAA	2-3 Wo	E
Pontocerebelläre Hypoplasie (PCH) * PCH2A, PCH2B, PCH2C, PCH4	P	277470 612389 612390 225753	TSEN54, TSEN2, TSEN34	2-4 Wo	E
Popliteales Pterygium-Syndrom (PPS)	P, M	119500 263650	IRF6, RIPK4	2-3 Wo	E
Prader-Willi-Syndrom (PWS) *, S	P, M, F	182279	15q11-q13-PWCR	2-4 Wo	E, FW, C
Prädisposition für fam. Brust- und Eierstockkrebs (BROVCA) *, S BROVCA1, BROVCA2, BROVCA3, BROVCA4, BROVCA Es erfolgt eine Stufendiagnostik gemäß EBM Kapitel 11.4.2 [GOP 11440].	P, M	604370 612555 613399 614291 114480	BRCA1, BRCA2, RAD51C, RAD51D, PLAB2, CHEK2	3-4 Wo	E
Prädisposition für Darmkrebs CRCS10, CRCS12	P	612591 615083	POLD1, POLE	2-4 Wo	E
Prädisposition für Gliome (GLM) GLM1, GLM2, GLM3, GLM9 *	P, M	137800 613028 613029 616568	TP53, PTEN, BRCA2, POT1	2-4 Wo	E
Prädisposition für Kutanes malignes Melanom (CMM) CMM1, CMM2, CMM3, CMM5, CMM10	P, M	155600 155601 609048 613099 615848	1p36-Chromosomenregion, CDKN2A, CDK4, MC1R, POT1	2-4 Wo	E
Prädisposition für nicht-kleinzelliges Lungenkarzinom (NSCLC)	P, M	211980	EGFR	2-4 Wo	E
Prädisposition für Pankreaskarzinom (PNCA) * PNCA2, PNCA3, PNCA4	P, M	260350 613347 614320	BRCA1, BRCA2, PALB2	3-4 Wo	E
Prädisposition für Prostatakrebs *	P, M	176807	CDH1, BRCA2, CHEK2, AR, HOXB13, HNF1B	2-3 Wo	E
Progressive externe Ophthalmoplegie (PEO) PEOA1, PEOB1	P, M	157640 258450	POLG	2-4 Wo	E
Propionazidämie	P, M, R	606054	PCCA, PCCB	2-4 Wo	E
Proteus-Syndrom *	P	176920	AKT1	2-3 Wo	E
Prune-Belly-Syndrom	P	100100	CHRM3	2-3 Wo	E
Pseudo-TORCH-Syndrom *	P	251290	OCLN	2-3 Wo	E, FW, C
Pseudo-von Willebrand-Krankheit (VWDP)	P	177820	GP1BA	2-3 Wo	E
Pseudoxanthoma elasticum (PXE) *	P, M	264800	ABCC6	3-4 Wo	E
Pyridoxamin-5'-Phosphat-Oxidase-Mangel	P	610090	PNPO	2-3 Wo	E
R					
Radioulnar-Synostose mit amegakaryozytärer Thrombozytopenie (RUSAT)	P	605432	HOXA11	2-4 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Retinitis pigmentosa (RP) RP1, RP2, RP3, RP4, RP7, RP19, RP23, RP37, RP38, RP39, RP42, RP43, RP44, RP51, RP54, RP56, RP74	P, M	268000 180100 312600 300029 613731 608133 601718 300424 611131 613862 613809 612943 613810 613769 613464 613428 613581 616562	RP1, RP2, RPGR, RHO, PRPH2, ABCA4, OFD1, NR2E3, MERTK, USH2A, KLHL7, PDE6A, RGR, TCC8, PCARE, IMPG2, BBS2	2-5 Wo	E
Restriktive Kardiomyopathie (RCM) * RCM1, RCM3, RCM4	P, M	115210 612422 615248	TNNI3, TNNT2, MYPN	2-4 Wo	E
Retinale Vaskulopathie mit zerebraler Leukodystrophie (RCVL)	P	192315	TREX1	2-3 Wo	E
Rett-Syndrom (RTT) RTT, Atypisches RTT; Kongenitales RTT	P, M	312750 613454	MECP2, FOXG1	2-4 Wo	E
Rhabdoid-Tumor-Prädispositionssyndrom 1 (RTPS1)	P, M	609322	SMARCB1	2-4 Wo	E
Rhizomele Chondrodysplasia punctata, Typ 1 (RCDP1)	P	215100	PEX7	2-4 Wo	E
Robinow-Sorauf-Syndrom	P, M	180750	TWIST1	2-3 Wo	E
Robinow-Syndrom RRS1, RRS2, A423, DRS1, DRS2	P, M	268310 818529 180700 616331	ROR2, NXN, WNT5A, DVL1	2-4 Wo	E
Rubinstein-Taybi-Syndrom (RSTS) RSTS1, RSTS2	P, M	180849 613684	CREBBP, EP300	3-4 Wo	E
S					
SADDAN-Dysplasie *	P	614482	FGFR3	2-3 Wo	E
Saethre-Chotzen-Syndrom (SCS)	P, M	101400	TWIST1	2-3 Wo	E, FW, C
Scapuloperoneale Myopathie, X-chromosomal, dominant (SPM)	P, M	300695	FHL1	2-3 Wo	E
Schilddrüsenkarzinom, familiäres medulläres (MTC)	P, M	155240	RET	2-3 Wo	E
Schinzel-Giedion-Syndrom (SGS)	P	269150	SETBP1	2-3 Wo	E
Schwannomatose (SWNTS) SWNTS1, SWNTS2	P, M	162091 615670	SMARCB1, NF2, LZTR1	2-4 Wo	E
Schwerhörigkeit, autosomal-rezessiv, 1 (DFNB1) *, S DFBN1A, DFBN1B Es erfolgt eine Stufendiagnostik gemäß EBM Kapitel 11.4.2.	P, M	220290 612645	GJB2, GJB6	2-3 Wo	E
Schwerhörigkeit, autosomal-rezessiv (DFNB) *, S DFNB1A, DFNB1B, DFNB4, DFNB18A, DFNB31, DFNB53, DFNB86, DFNB97 Es erfolgt eine Stufendiagnostik gemäß EBM Kapitel 11.4.2	P, M	220290 612645 600791 602092 607084 609706 614617 616705	GJB2, GJB6, SLC26A4, USH1C, WHRN, MET, TBC1D24, COL11A2	2-5 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Schwerhörigkeit, autosomal-dominant (DFNA) * DFNA2B, DFNA3A, DFNA3B, DFNA5, DFNA13, DFNA17, DFNA65, DFNA34	P, M	612644 601544 612643 600994 601868 603622 616044 617772	GJB3, GJB2, GJB6, GSDME, COL11A2, MYH9, NLRP, TBC1D24	2-5 Wo	E
Seckel-Syndrom (SCKL) SCKL1, SCKL4, SCKL5	P, M	210600 613676 613823	ATR, CENPJ, CEP152	2-4 Wo	E
Sensorisch-ataktische Neuropathie, Dysarthrie und Ophthalmoparese (SANDO)	P, M	607459	POLG	2-4 Wo	E
Septo-optische Dysplasie	P	182230	HESX1	2-3 Wo	E
Shwachman-Diamond-Syndrom (SDS)	P	260400	SBDS	2-3 Wo	E
Short-QT-Syndrom (SQT) * SQT1, SQT2, SQT3	P, M	609620 609621 609622	KCNH2, KCNQ1, KCNJ2	2-4 Wo	E
SHORT-Syndrom	P	269880	PIK3R1	2-3 Wo	E
Sichelzellenanämie *	P	603903	HBB	2 Wo	E, FW, C
Sick-Sinus-Syndrom (SSS) SSS1, SSS2, SSS3	P	608567 163800 614090	SCN5A, HCN4, MYH6	2-4 Wo	E
Silver-Russell-Syndrom (SRS) *	F, M	180860	11p15-ICR1, Chromosom-7-Region	2-4 Wo	E
Simpson-Golabi-Behmel-Syndrom (SGBS) SGBS1, SGBS2	P, M	312870 300209	GPC3, GPC4, OFD1 (CXORF5)	2-4 Wo	E
Sitosterolämie (STSL)	P	210250	ABCG8, ABCG5	2-3 Wo	E
Skewed X-Inaktivierung, familiärer Typ 1 (SXI1) S	P, F	300087	XIST	2-3 Wo	E
Smith-Lemli-Opitz-Syndrom (SLOS)	P	270400	DHCR7	2-3 Wo	E, FW, C
Smith-Magenis-Syndrom (SMS) S	P, M	182290 610882	17p11.2-SMCR, RAI1	2-4 Wo	E, FW, C
Smith-McCort-Dysplasie (SMC) * SMC1, SMC2	P	607326 615222	DYM, RAB33B	2-3 Wo	E, FW, C
Sotos-Syndrom (SOTOS) SOTOS1, SOTOS2, SOTOS3	P, M	117550 614753 617169	NSD1, NFIX, APC2	2-4 Wo	E
Spalthand-Spaltfuß-Malformation 6 (SHFM6)	P	225300	WNT10B	2-3 Wo	E
Spastische Paraplegie (SPG) SPG1, SPG2, SPG4, SPG15, SPG17, SPG23, SPG39, SPG43, SPG44	P, M	303350 312920 182601 270700 270685 270750 612020 615043 613206	L1CAM, PLP1, SPAST, ZFYVE26, BSCL2, DSTYK, PNPLA6, C19ORF12, GJC2	2-4 Wo	E
Störung der Spermatogenese, Y-chromosomal (SPGFY) * SPGFY1, SPGFY2	P, M, F	400042 415000	AZF	2-3 Wo	E
Spinale Muskelatrophie (SMA) * SMA1, SMA2, SMA3, SMA4	M	253300 253550 253400 271150	SMN1, SMN2	2-3 Wo	E, FW, C
Spinale Muskelatrophie, distale (DSMA) DSMA1, DSMAX	P, M	604320 300489	IGHMBP2, ATP7A	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Spinale und bulbäre Muskelatrophie 1, X-chromosomal (SMAX1)	F, B	313200	AR	2-3 Wo	E, FW, C
Spondyloepiphysäre Dysplasie (SED) SEDC, SEDSTN, SED, Maroteaux-Typ	P, M	183900 616583 184095	COL2A1, TRPV4	3-5 Wo	E
Sprech- und Sprachstörung, Typ 1 (SPCH1)	P, M	602081	FOXP2	2-3 Wo	E
Statin-Unverträglichkeit [CO]	P	604843	SLCO1B1-Polymorphismus	2-3 Wo	E
Stargardt-Krankheit (STGD) STGD1, STGD3	P, M	248200 600110	ABCA4, CNGB3, ELOVL4	2-4 Wo	E
Stickler-Syndrom (STL) STL1, STL2	P, M	108300 609508 604841	COL2A1, COL11A1	3-5 Wo	E
T					
Tangier-Krankheit (TGD)	P	205400	ABCA1	2-3 Wo	E
Teleangiektasie, hereditäre hämorrhagische (HHT) HHT1, HHT2, HHT5	P, M	187300 600376 615506	ENG, ACVRL1, GDF2	2-4 Wo	E
Thanatophore Dysplasie (TD) [*, S] TD1, TD2	P	187600 187601	FGFR3	2-4 Wo	E, FW, C
Thiamin-Metabolismus-Dysfunktionssyndrom 5 (THMD5)	P	614458	TPK1	2-4 Wo	E
Timothy-Syndrom (TS) [*]	P	601005	CACNA1C	2-3 Wo	E
Thorakales Aortenaneurysma (AAT) *, S AAT3, AAT4, AAT5, AAT6, AAT7, AAT11, LDS3, LDS4, LDS5, MFS, EDVASC	P	610168 132900 609192 611788 613780 617349 613795 614816 154700 130050	TGFBR2, MYH11, TGFBR1, ACTA2, MYLK, FOXE3, SMAD3, TGFB2, TGFB3, FBN1, COL3A1	2-5 Wo	E
Thrombophilie [*, CO] THPH1, THPH2, THPH3, THPH4, THPH5, THPH6, THPH7	P, M, S	188050 188055 176860 612304 612336 614514 613118	F2, F5, PROC, PROS1, SERPINC1	2-4 Wo	E
Thrombozytenbedingte Blutungsneigung (BDPLT) BDPLT1, BDPLT2, BDPLT3, BDPLT4, BDPLT6, BDPLT9, BDPLT16	P	231200 273800 177820 139090 155100 614200 187800	GP1BA, GP1BB, GP9, ITGA2B, ITGB3, NBEAL2, MYH9, ITGA2, ITGA2B, ITGB3	2-5 Wo	E
Transiente neonatale Hyperbilirubinämie, familiäre (HBLRTFN)	P	237900	UGT1A1	2-3 Wo	E
Transthyretin-Amyloidose, hereditäre	P	105210	TTR	2-3 Wo	e
Treacher-Collins-Syndrom (TCS) TCS1, TCS2, TCS3	P, M	154500 613717 248390	TCOF1, POLR1D, POLR1C	3-4 Wo	E
Trichothiodystrophie 1, nicht-photosensitive (TTDN1)	P	234050	MPLKIP	2-3 Wo	E
Tuberöse Sklerose (TSC) TSC1, TSC2	P, M	191100 613254	TSC1, TSC2	3-4 Wo	E, FW, C

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
U					
Usher-Syndrom (USH) USH1C, USH2A, USH2C, USH2D	P, M	276904 276901 605472 611383	USH1C, USH2A, ADGRV1, PDZD7, WHRN	2-5 Wo	E
V					
Van der Woude-Syndrom 1 (VWS1)	P, M	119300	IRF6	2-3 Wo	E
Variables Aneuploidie-Mosaik-Syndrom 1 (MVA1)	P	257300	BUB1B	2-4 Wo	E
Velokardiofaziales Syndrom (VCFS) [S]	P, M	192430	22q11.2-DGCR, TBX1	2-3 Wo	E, FW, C
Venöse Malformation, multiple kutane und muköse (VMCM)	P	600195	TEK	3-4 Wo	E
Ventrikelseptumdefekt (VSD) VSD1, VSD2	P	614429 614432	GATA4, NKX2-5	2-3 Wo	E
Ventrikuläre Tachykardie, katecholaminerge polymorphe (CPVT) CPVT1, CPVT2	P	604772 611938	RYR2, CASQ2	3-4 Wo	E
Vohwinkel-Syndrom *	P	124500	GJB2	2-3 Wo	E
Von-Hippel-Lindau-Syndrom *	P, M	193300	VHL	2-3 Wo	E
Vorhofflimmern, familiäres (ATFB) * ATFB3, ATFB4, ATFB9, ATFB10, ATFB13, ATFB16	P, M	607554 611493 613980 614022 615377 613120	KCNQ1, KCNE2, KCNJ2, SCN5A, SCN1B, SCN3B	2-4 Wo	E
Vorzeitige Ovarialinsuffizienz (POF) POF1, POF2B, POF3, POF4, POF7, POF9, POF10	P, M	311360 300604 608996 300510 612964 615724 612885	FMR1, POF1B, FOXL2, BMP15, NR5A1, HFM1, MCM8	2-4 Wo	E
W					
Waardenburg-Syndrom (WS) WS1, WS2D, WS2E, WS3, WS4C	P, M	193500 608890 611584 148820 613266	PAX3, SNAI2, SOX10	2-4 Wo	E
Wachstumshormon-Insensitivität, partielle (GHIP)	P, M	604271	GHR	2-3 Wo	E
Walker-Warburg-Syndrom (WWS, MDDGA) MDDGA4, MDDGA5, MDDGA7, MDDGA11	P	253800 613153 614643 615181	FKTN, FKRP, ISPD, B3GALNT2	2-4 Wo	E
Warburg-Mikro-Syndrom (WARBM1)	P	600118	RAB3GAP1	3-4 Wo	E, FW, C
Watson-Syndrom (WTSN) *	P, M	193520	NF1	3-4 Wo	E
Weill-Marchesani-Syndrom 2 (WMS2) *	P, M	608328	FBN1	3-6 Wo	E
Williams-Beuren-Syndrom (WBS)	M	194050 609757 613729	7q11.23-Chromosomenregion	2-3 Wo	E, FW, C
Wilms-Tumor (WT) WT1, WT2	P, M	194070 194071	WT1, BRCA2, ICR1-Chromosomenregion	2-4 Wo	E
Wilson-Krankheit (WD) *	P, M	277900	ATP7B	2-4 Wo	E
Wolff-Parkinson-White-Syndrom (WPW) *	P	194200	PRKAG2	2-3 Wo	E
Wolf-Hirschhorn-Syndrom (WHS)	M	194190	4p16.3-Chromosomenregion	2-3 Wo	E, FW, C
Wolman-Krankheit	P	278000	LIPA	2-3 Wo	E

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Z					
Zahnagenesie, selektive (STHAG) STHAG1, STAHG4, STAHG9, STHAGX1	P, M	106600 150400 617275 313500	MSX1, WNT10A, GREM2, EDA	2-4 Wo	E
Zapfen-Stäbchen-Dystrophie (CORD) CORDX1, CORD3	P, M	304020 604116	RPGR, ABCA4	2-4 Wo	E
Zentrales Hypoventilationssyndrom, kongenitales (CCHS) [S]	P, M, F	209880	PHOX2B	2-3 Wo	E
Zentronukleäre Myopathie, X-chromosomal (CNMX)	P	310400	MTM1	2-3 Wo	E
Zerebrale kavernöse Fehlbildung (CCM) CCM1, CCM2, CCM3	P, M	116860 603284 603285	KRIT1, CCM2, PDCD10	2-4 Wo	E
Zerebrales Kreatinmangel-Syndrom (CCDS) CCDS1, CCDS2	P	300352 612736	SLC6A8, GAMT	2-4 Wo	E
Ziliendyskinesie, primäre (CILD) * CILD1, CILD3, CILD7, CILD	P, M	244400 608644 611884	DNAI1, DNAH5, DNAH11, DNAH8	3-6 Wo	E, FW, C
Zystische Fibrose (CF) *, [S]	N, M, P	219700	CFTR	2-4 Wo	E, FW, C
3					
3M-Syndrom 3M1, 3M2	P	273750 612921	CUL7, OBSL1	2-4 Wo	E
4					
46,XX-Geschlechtsumkehr (SRXX) SRXX1, SRXX4	P, M	400045 278850	SRY, NR5A1	2-3 Wo	E, FW, C
46,XY-Geschlechtsumkehr (SRXY) SRXY1, SRXY2, SRXY3, SRXY7, SRXY9	P, M	400044 300019 612965 233420 616067	SRY, NR0B1, NR5A1, DHH, ZFPM	2-3 Wo	E, FW, C
Uniparentale Disomie (UPD)					
Untersuchungsmaterial: E/FW/C vom Patient und E von beiden Eltern					
Uniparentale Disomie 7 (matUPD7) * Silver-Russel-Syndrom (RSS)	F, M	180860	7q32-CR	2-4 Wo	E, FW, C
Uniparentale Disomie 11 (patUPD11) * Beckwith-Wiedemann-Syndrom (BWS)	F, M	130650	11p15-BWCR	2-4 Wo	E, FW, C
Uniparentale Disomie 14 (patUPD14) * Kagami-Ogata-Syndrom	F, M	608149	14q32-CR	2-4 Wo	E, FW, C
Uniparentale Disomie 15 (matUPD15) * Prader-Willi-Syndrom (PWS)	F, M	176270	15q11-q13-PWCR	2-4 Wo	E, FW, C
Uniparentale Disomie 15 (patUPD15) * Angelman-Syndrom (AS)	F, M	105830	15q11-q13-ANCR	2-4 Wo	E, FW, C

Erkrankung / Diagnostik	Methode	OMIM	Gen/Chromosomenregion	Dauer	Material
Ausschluss mütterlicher Kontamination					
Untersuchungsmaterial: 2 ml EDTA-Blut der Schwangeren					
AMK * Im Rahmen jeder pränatalen molekularetischen Diagnostik ist eine Mikrosatellitenanalyse mit mütterlichem EDTA-Blut zum Ausschluss einer maternalen Kontamination notwendig.	F			1-2 T	E
Pränataler Schnelltest					
Pränataler Schnelltest (AnEu-PCR) * (Chromosom 21, 18, 13, X und Y)	F			1-2 T	FW, C, A
Postnataler Schnelltest					
Postnataler Schnelltest (AnEu-PCR) * (Chromosom 21, 18, 13, X und Y)	F			1-2 T	E
Hinweise					
* = akkreditiertes Verfahren					
co = in Kooperation					
S = Stufendiagnostik					
Methodenkürzel					
PCR + Sequenzierung	P				
MLPA	M				
NGS	N				
Mikrosatelliteninstabilität	MSI				
Immunhistochemie	I				
ARMS (Amplification-refractory mutation system)	AR				
Fragmentanalyse	F				
Methylierungsanalyse	MA				
Blot	B				
Array-CGH	A				
SNP-Analyse (TaqMan)	S				
Real-Time-PCR-Analyse (qPCR)	R				
Exon-Quantifizierung mittels NGS-Reads-Statistik	E				