SYNLAB MVZ Humangenetik Mannheim

Stand: 01.08.2025

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".**

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Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Achromatopsie (ACHM) *	3 - 5 Wo	E
Gen-Panel: ID164.02, 6 Gene (10,4 kb)		
ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H		
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		
Altersbedingte Makuladegeneration (AMD, ARMD) *	3 - 5 Wo	E
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		
Anophthalmie und Mikrophthalmie (MCOP) *	4 - 6 Wo	E
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		
Dysgenesie des vorderen Augensegmentes (ASGD) *	3 - 5 Wo	E
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		
Fraser-Syndrom (FRASRS) *	3 - 5 Wo	E
Gen-Panel: ID317.00, 3 Gene (24,8 kb)		
FRAS1, FREM2, GRIP1		
Fuchs-Endotheldystrophie (FECD) *	3 - 5 Wo	E
Gen-Panel: ID261.00, 5 Gene (13,5 kb)		
AGBL1, COL8A2, SLC4A11, TCF4, ZEB1		
Glaukom (GLC) *	3 - 5 Wo	E
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		
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		
Hohe Myopie (MYP) *	3 - 5 Wo	E
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		

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
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) 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	3 - 5 Wo	E
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) 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	3 - 5 Wo	E
Kongenitale extraokuläre Muskelfibrose (CFEOM) * Gen-Panel: ID063.00, 5 Gene (10,5 kb) COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3	3 - 5 Wo	E
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 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	3 - 5 Wo	E
Kongenitale stationäre Nachtblindheit (CSNB) * Gen-Panel: ID267.00, 14 Gene (39,2 kb) CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, SAG, RHO, SLC24A1, TRPM1	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
Lebersche kongenitale Amaurose (LCA) * Gen-Panel: ID187.01, 19 Gene (38,8 kb) AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45	3 - 5 Wo	E
Makuladystrophie (MD) * Gen-Panel: ID139.02, 18 Gene (44,6 kb) ABCA4, BEST1, CDH3, CHST6, CNGB3, CRB1, CTNNA1, ELOVL4, IMPG1, IMPG2, MAPKAPK3, MFSD8, PROM1, PRPH2, RDH12, RP1L1, SIX6, TIMP3	3 - 5 Wo	E
Morbus Stargardt (STGD) * Gen-Panel: ID102.01, 4 Gene (11,4 kb) ABCA4, ELOVL4, PROM1, PRPH2	3 - 5 Wo	E
Optikusatrophie (OPA) * 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	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Okulokutaner Albinismus (OCA) * Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E
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
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
Retinitis pigmentosa (RP), autosomal-dominant * 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	3 - 5 Wo	E
Retinitis pigmentosa (RP), autosomal-rezessiv * 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	4 - 6 Wo	E
Retinitis pigmentosa (RP), umfassende Diagnostik * 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	4 - 6 Wo	E
Senior-Loken-Syndrom (SLSN) * Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E
Septooptische Dysplasie * Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
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
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 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	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
Weill-Marchesani-Syndrom (WMS) * Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD) * Gen-Panel: ID101.03, 38 Gene (94,5 kb) 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	3 - 5 Wo	E
Bindegewebserkrankungen		
Bindegewebserkrankungen mit Aortenbeteiligung * Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, TNXB, ZNF469 Thorakales Aortenaneurysma und Aortendissektion (TAA/D): 17 Gene (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFBR2, TGFBR1 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
Ehlers-Danlos-Syndrom (EDS) * Gen-Panel: ID039.05 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 8 Gene (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1 Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	3 - 5 Wo	E
Hereditäre hämorrhagische Teleangiektasie (HHT) * Gen-Panel: ID155.01, 7 Gene (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Bindegewebserkrankungen		
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E
Marfan-Syndrom (MFS) * Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2	2 - 4 Wo	E
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.05 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 48 Gene (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, 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-Syndrom (LDS) und Thorakales Aortenaneurysma (AAT): 17 Gene (42,6 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 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	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) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) * Gen-Panel: ID020.01 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 16 Gene (41,0 kb) ACTA2, COL3A1, FBN1, FOXE3, 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

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
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
Hyperparathyreoidismus (HRPT) * Gen-Panel: ID338.00, 10 Gene (19,2 kb) AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6	3 - 5 Wo	E
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung * 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	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
Hypoparathyreoidismus * 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	3 - 5 Wo	E
Hypophysenadenom (PITA) * Gen-Panel: ID387.01, 21 Gene (47,3 kb) AIP, CDH23, CDKN1B, DICER1, EPCAM, GNAS, GPR101, MAX, MLH1, MSH2, MSH6, MEN1, PMS2, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, USP8	3 - 5 Wo	E
Kongenitale Hypothyreose * 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 (23,9 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	3 - 5 w	E
MODY-Diabetes * Gen-Panel: ID048.01, 14 Gene (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 Wo	E
Neuroendokrine Neoplasie * 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	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
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
Phäochromozytom-Paragangliom-Syndrom (PPGL) * Gen-Panel: ID042.03, 14 Gene (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E
Primäre und prämature Ovarialinsuffizienz (POI, POF) * 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	3 - 5 Wo	E
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) * 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	3 - 5 Wo	E
Schilddrüsenkarzinom * 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	3 - 5 Wo	E
Septooptische Dysplasie * Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
Störung der Geschlechtsentwicklung (DSD) * Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSPO1, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX9, 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

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Adipositas *	4 - 6 Wo	E
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		
Autismus-Spektrum-Störungen *	4 - 6 Wo	E
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		
Coffin-Siris-Syndrom (CSS) *	3 - 5 Wo	E
Gen-Panel: ID118.01, 14 Gene (46,5 kb)		
ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11		
Cornelia-de-Lange-Syndrom (CDLS) *	3 - 5 Wo	E
Gen-Panel: ID033.02, 8 Gene (32,0 kb)		
ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS		
FG-Syndrom (FGS) *	3 - 5 Wo	E
Gen-Panel: ID215.00, 3 Gene (17,2 kb)		
CASK, FLNA, MED12		
Großwuchs, umfassende Diagnostik *	4 - 6 Wo	E
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		
Großwuchssyndrome (SOTOS, BWS) *	3 - 5 Wo	E
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
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, LING01, 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, LING01, 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		
Neurologische Entwicklungsstörungen (NED), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID358.00, 169 Gene (431,8 kb)		
ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDFT1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNPK, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, 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, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6		

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Kabuki-Syndrom (KABUK) *	3 - 5 Wo	E
Gen-Panel: ID127.00, 2 Gene (20,8 kb)		
KDM6A, KMT2D		
Kleinwuchs, umfassende Diagnostik *	4 - 6 Wo	E
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		
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		
Pitt-Hopkins-Syndrom (PTHS) *	3 - 5 Wo	E
Gen-Panel: ID106.00, 3 Gene (10,6 kb)		
CNTNAP2, NRXN1, TCF4		
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		
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		
Rett-Syndrom (RTT) und ähnliche Krankheitsbilder *	3 - 5 Wo	E
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		
Rubinstein-Taybi-Syndrom (RSTS) *	3 - 5 Wo	E
Gen-Panel: ID142.01, 3 Gene (24,3 kb)		
CREBBP, EP300, SRCAP		
Seckel-Syndrom (SCKL) *	3 - 5 Wo	E
Gen-Panel: ID113.00, 9 Gene (33,5 kb)		
ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP		
Sprachentwicklungsstörung (DLD, CAS) *	4 - 6 Wo	E
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		

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Wachstumshormonmangel (IGHD, CPHD) *	3 - 5 Wo	E
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		
Wachstumsstörung und Makrozephalie *	3 - 5 Wo	E
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		
3M-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID214.00, 3 Gene (12,4 kb)		
CCDC8, CUL7, OBSL1		
Epilepsien und Migräne		
Absence-Epilepsie (EJA, ECA) *	3 - 5 Wo	E
Gen-Panel: ID057.02, 10 Gene (21,8 kb)		
CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, KCNMA1, RORB, SLC2A1, SLC12A5		
Benigne neonatale und infantile Krampfanfälle (BFNS, BFIS) *	3 - 5 Wo	E
Gen-Panel: ID134.01, 6 Gene (19,8 kb)		
CHRNA2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A		
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE) *	4 - 6 Wo	E
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		
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Epilepsien und Migräne		
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, SLC7A6OS, 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) 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, SLC7A6OS, 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	4 - 6 Wo	E
Epilepsie mit schwerer Entwicklungsstörung * 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	3 - 5 Wo	E
Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFSP) * 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	3 - 5 Wo	E
Fokale Epilepsien * 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	3 - 5 Wo	E
Generalisierte Epilepsien * 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, SLC7A6OS, 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, SLC7A6OS, 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	3 - 5 Wo	E
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFSP, GEFS+) * Gen-Panel: ID235.03, 8 Gene (17,2 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, SLC32A1, STX1B	3 - 5 Wo	E
Hemiplegische Migräne (FHM) * Gen-Panel: ID064.02, 7 Gene (23,1 kb) ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Epilepsien und Migräne		
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
Migräne (MGR) * 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	3 - 5 Wo	E
Myoklonusepilepsie (EPM, EJM) * 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	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
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
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: ID391.00, 60 Gene (141,6 kb) ADA2, 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
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
Fertilitätsstörungen		
Männliche Infertilität, umfassende Diagnostik * Gen-Panel ID192.06 Männliche Infertilität, umfassende Diagnostik: 126 Gene (424,9 kb) ACR, ACTL7A, ACTL9, ADA2, ADGRG2, AK7, AK9, AKAP3, AR, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRRC23, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RAD21L1, RBBP7, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SPO11, SSX1, STAG3, STK33, STRA8, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Störung der Spermatogenese (SPGF): 107 Gene (368,8 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CT55, CYLC1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, 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, 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
Ovarialdysgenesie (ODG) * 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	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
Primäre und prämature Ovarialinsuffizienz (POI, POF) * 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	3 - 5 Wo	E
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) * 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	4 - 6 Wo	E
Störung der Geschlechtsentwicklung (DSD) * Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSPO1, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX9, 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
Störung der Oozyten-, Zygoten- und Embryonen-Reifung (OZEMA) * 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	3 - 5 Wo	E
Zystische Fibrose (CF) * Gen-Panel: ID045.00, 1 Gen (4,4 kb) CFTR	2 - 4 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Fertilitätsstörungen		
Weibliche Infertilität, umfassende Diagnostik * 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	3 - 5 Wo	E
Fettstoffwechselstörungen		
Fettstoffwechselstörungen, umfassende Diagnostik * 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	4 - 6 Wo	E
Fettstoffwechselstörung durch LDL-Mangel * Gen-Panel: ID094.01, 10 Gene (29,6 kb) ANGPTL3, ANGPTL4, ANGPTL8, APOB, APOE, LIMA1, MTTP, NPC1L1, PCSK9, SAR1B	3 - 5 Wo	E
HDL-assoziierte Fettstoffwechselstörung * Gen-Panel: ID096.01, 15 Gene (25,5 kb) ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA2, APOA4, APOC3, CETP, LCAT, LIPC, LIPG, NPC1, NPC2, SCARB1, SMPD1	3 - 5 Wo	E
Hypertriglyzeridämie * 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	3 - 5 Wo	E
Hypercholesterinämie * Gen-Panel: ID024.02, 13 Gene (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1	3 - 5 Wo	E
Kombinierte Hyperlipidämie * 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	3 - 5 Wo	E
Lipodystrophie (CGL, FPLD) * 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	3 - 5 Wo	E
Lipodystrophien, umfassende Diagnostik * 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	3 - 5 Wo	E
Statin-assoziierte Myopathie * Gen-Panel: ID097.00, 11 Gene (37,2 kb) ACADM, ACADS, ACADVL, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1, SLCO1B1	3 - 5 Wo	E

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		
Lynch-Syndrom (LYNCH, HNPCC) *	3 - 5 Wo	E
Gen-Panel: ID002.02, 5 Gene (12,7 kb)		
MLH1, MSH2, MSH6, PMS2, EPCAM		
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 und Polyposis *	3 - 5 Wo	E
Gen-Panel ID006.09, 23 Gene (64,1 kb)		
APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53		
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: ID005.06, 15 Gene (39,4 kb)		
APC, AXIN2, 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, TOGARAM1, 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: ID392.00 Anämien, umfassende Diagnostik: 187 Gene (356,5 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, 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): 2 Gene (11,1 kb) HEATR3, RPL5, RPL9, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPS7, RPS10, RPS15A, 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
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
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
Hämatologische Erkrankungen		
Knochenmarkinsuffizienz (BMF) *	3 - 5 Wo	E
Gen-Panel: ID357.00		
Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb):		
ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTEL1, SRP72, TERC, TERT, TP53, ZCCHC8		
Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb)		
ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53		
Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb)		
PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Neutropenie *	3 - 5 Wo	E
Gen-Panel: ID189.02		
Neutropenie: 30 Gene (51,0 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1		
Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS		
Syndrome mit Neutropenie: 21 Gene (37,5 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, 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		
Thrombozytendefekte, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID274.01		
Thrombozytendefekte, umfassende Diagnostik: 64 Gene (142,3 kb)		
ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, EPHB2, ETV6, FERMT3, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB3, JAK2, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TUBB1, WAS, WDR1, WIPF1		
Thrombozytopenie, nicht-syndromal (THC): 9 Gene (17,7 kb)		
ANKRD26, CYCS, ETV6, FYB1, IKZF5, MASTL, SRC, TUBB1, WAS		
Thrombozythämie (THCYT): 3 Gene (6,4 kb)		
JAK2, MPL, THPO		
Thrombozytenbedingte Blutungsstörung (BDPLT): 23 Gene (52,8 kb)		
ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1		
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 (THC) *	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: ID109.02, 11 Gene (27,9 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
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 (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		
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, C30RF52, 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		
Syndromale 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		

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Gorlin-Syndrom (BCNS) und ähnliche Krankheitsbilder * Gen-Panel: ID174.02, 8 Gene (25,8 kb) BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 Wo	E
Hereditäres Angioödem (HAE) * Gen-Panel: ID345.00, 7 Gene (16,4 kb) ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1	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
Hydrops fetalis * Gen-Panel: ID370.00, 148 Gene (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	3 - 5 w	E
Hypotrichose, nicht-syndromale Form (HYPT) * Gen-Panel: ID146.01, 12 Gene (20,1 kb) APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE	3 - 5 Wo	E
Ichthyose * 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	3 - 5 Wo	E
Kutanes malignes Melanom (CMM) * Gen-Panel: ID193.01, 12 Gene (26,4 kb) BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3	3 - 5 Wo	E
Multiples Pterygium-Syndrom * Gen-Panel: ID158.01, 8 Gene (16,8 kb) CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	3 - 5 Wo	E
Neurofibromatose (NF) * Gen-Panel: ID210.00, 3 Gene (11,6 kb) NF1, NF2, SPRED1	3 - 5 Wo	E
Okulokutaner Albinismus (OCA) * Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E
Orofaziodigitales Syndrom (OFD) * Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E
Pachyonychia congenita (PC) * Gen-Panel: ID120.01, 13 Gene (30,1 kb) AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3	3 - 5 Wo	E
Palmoplantarkeratose (PPK) * 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	3 - 5 Wo	E
Peeling-Skin-Syndrom (PSS) * Gen-Panel: ID309.00, 6 Gene (13,6 kb) CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5	3 - 5 Wo	E
Pierre-Robin-Sequenz * Gen-Panel: ID294.00, 33 Gene (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Primäres Lymphödem (LMPHM) * 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	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
Selektive Zahn-Agenesie (STHAG) * 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	3 - 5 Wo	E
Systemerkrankungen mit multiplen Café-au-lait-Flecken * 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	3 - 5 Wo	E
Tuberöse Sklerose (TSC) * Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
Xeroderma pigmentosum (XP) * Gen-Panel: ID282.00, 10 Gene (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 Wo	E
Herz- und Gefäßerkrankungen		
Alagille-Syndrom (ALGS) * Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC) * 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	4 - 6 Wo	E
Atriumseptumdefekt und Ventrikelseptumdefekt (ASD, VSD, AVSD) * Gen-Panel: ID143.00, 12 Gene (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1	3 - 5 Wo	E
Bikuspide Aortenklappe (AOVD) * Gen-Panel: ID301.00, 6 Gene (16,7 kb) GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2	3 - 5 Wo	E
Bindegewebserkrankungen mit Aortenbeteiligung * Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, TNXB, ZNF469 Thorakales Aortenaneurysma und Aortendissektion (TAA/D): 17 Gene (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFBR2, TGFBR1 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
Brugada-Syndrom (BRGDA) * 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	3 - 5 Wo	E
CADASIL und CARASIL * Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
CHARGE-Syndrom * Gen-Panel: ID307.00, 3 Gene (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 Wo	E
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
Dilatative Kardiomyopathie (CMD, DCM) * 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	4 - 6 Wo	E
Ehlers-Danlos-Syndrom (EDS) * Gen-Panel: ID039.05 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 8 Gene (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1 Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	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
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
Familiäres 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
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
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

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Hereditäre hämorrhagische Teleangiektasie (HHT) *	3 - 5 Wo	E
Gen-Panel: ID155.01, 7 Gene (15,6 kb)		
ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4		
Herz-Hand-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID165.01, 9 Gene (23,8 kb)		
DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5		
Hypertrophe Kardiomyopathie (CMH, HCM) *	4 - 6 Wo	E
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		
Kardiale Arrhythmien, umfassende Diagnostik *	4 - 6 Wo	E
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		
Kardiomyopathien, umfassende Diagnostik *	4 - 6 Wo	E
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		
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) *	3 - 5 Wo	E
Gen-Panel: ID012.03, 9 Gene (33,9 kb)		
ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN		
Konotrunkale Herzfehlbildung (CTHM) *	3 - 5 Wo	E
Gen-Panel: ID160.01, 12 Gene (24,8 kb)		
CFC1, FLT4, FOXH1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3		

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
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
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, 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
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.05 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 48 Gene (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, 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-Syndrom (LDS) und Thorakales Aortenaneurysma (AAT): 17 Gene (42,6 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 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	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

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
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, PPH) * 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
Syndromale kongenitale Herzfehler * Gen-Panel ID252.02 Synromale 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

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Short-QT-Syndrom (SQT) *	3 - 5 Wo	E
Gen-Panel: ID233.01, 7 Gene (22,1 kb)		
CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3		
Sick-Sinus-Syndrom (SSS) *	3 - 5 Wo	E
Gen-Panel: ID107.01, 4 Gene (16,5 kb)		
GNB2, HCN4, MYH6, SCN5A		
Speicherkrankheiten mit Herzbeteiligung *	3 - 5 Wo	E
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		
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) *	3 - 5 Wo	E
Gen-Panel: ID020.01		
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 16 Gene (41,0 kb)		
ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Nicht-syndromales thorakales Aortenaneurysma (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		
Viszerale Heterotaxie (HTX) *	3 - 5 Wo	E
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		
Zerebrale Kleingefäßerkrankung (BSVD) *	3 - 5 Wo	E
Gen-Panel: ID325.00, 3 Gene (12,0 kb)		
COL4A1, COL4A2, COLGALT1		
Zerebrovaskuläre Erkrankungen und Schlaganfall *	4 - 6 Wo	E
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		
HNO-Erkrankungen		
Branchiootorenales Syndrom (BOR) *	3 - 5 Wo	E
Gen-Panel: ID315.00, 5 Gene (10,2 kb)		
EYA1, SALL1, SIX1, SIX5, TFAP2A		
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		
Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX) *	3 - 5 Wo	E
Gen-Panel: ID290.01, 8 Gene (17,1 kb)		
AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX, TIMM8A		
Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB) *	4 - 6 Wo	E
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		

Erkrankung/Diagnostik	Dauer	Material
HNO-Erkrankungen		
Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA) *	4 - 6 Wo	E
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		
Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik *	4 - 6 Wo	E
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		
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		
Syndromale Schwerhörigkeit, umfassende Diagnostik *	4 - 6 Wo	E
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		
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
Immunologische Erkrankungen		
Antikörpermangelsyndrome (CVID, AGM) *	3 - 5 Wo	E
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		
Atypisches hämolytisch-urämisches Syndrom (AHUS) *	3 - 5 Wo	E
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		
Autoinflammatorische Erkrankungen, umfassende Diagnostik *	3 - 5 Wo	E
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		
Chronische Granulomatose (CGD) *	3 - 5 Wo	E
Gen-Panel: ID379.00, 8 Gene (8,8 kb)		
CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2		
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		
Hyper-IgE-Syndrom mit rekurrenten Infektionen (HIES) *	3 - 5 Wo	E
Gen-Panel: ID240.01, 10 Gene (29,5 kb)		
DOCK8, DSG1, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TYK2, ZNF341		
Knochenmarkinsuffizienz (BMF) *	3 - 5 Wo	E
Gen-Panel: ID357.00		
Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb):		
ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTEL1, SRP72, TERC, TERT, TP53, ZCCHC8		
Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb)		
ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53		
Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb)		
PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8		
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, C2ORF69, 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		

Erkrankung/Diagnostik	Dauer	Material
Immunologische Erkrankungen		
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
Neutropenie * Gen-Panel: ID189.02 Neutropenie: 30 Gene (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndrome mit Neutropenie: 21 Gene (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, USB1, VPS13B, WDR1	3 - 5 Wo	E
Schwerer kombinierter Immundefekt (SCID) * 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	3 - 5 Wo	E
Thrombotische Mikroangiopathie (TMA) * 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,	3 - 5 Wo	E
Lungen- und Respiratorische Erkrankungen		
Chronische Granulomatose (CGD) * Gen-Panel: ID379.00, 8 Gene (8,8 kb) CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2	3 - 5 Wo	E
Interstitielle Lungenerkrankung und Lungenfibrose * 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	3 - 5 Wo	E
Kongenitales zentrales Hypoventilationssyndrom (CCHS) * Gen-Panel: ID185.01, 5 Gene (9,7 kb) LBX1, MECP2, MYO1H, PHOX2B, RET	2 - 4 Wo	E
Lungenkarzinom * 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	4 - 6 Wo	E
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) * 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	4 - 6 Wo	E
Pulmonale Hypertonie (PAH, PPH) * 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
Pulmonale Surfactant-Stoffwechselstörung (SMDP) * Gen-Panel: ID168.01, 6 Gene (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 Wo	E
Zystische Fibrose (CF) * Gen-Panel: ID045.00, 1 Gen (4,4 kb) CFTR	2 - 4 Wo	E

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		
Hydrozephalus *	3 - 5 Wo	E
Gen-Panel: ID221.04		
Hydrozephalus: 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
Mikrozephalien und Makrozephalien		
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		
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

Erkrankung/Diagnostik	Dauer	Material
Mitochondriopathien		
Coenzym-Q10-Mangel (COQ10D) *	3 - 5 Wo	E
Gen-Panel: ID225.01, 15 Gene (18,7 kb)		
ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2		
Kombinierter Defekt der oxidativen Phosphorylierung (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		
Lebersche hereditäre Optikusneuropathie (LHON) [CO]	3 - 5 Wo	E
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		
MELAS-Syndrom [CO]	3 - 5 Wo	E
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		
Mitochondriales DNA-Depletionssyndrom (MTDPS) *	3 - 5 Wo	E
Gen-Panel: ID324.00, 19 Gene (25,6 kb)		
AGK, DGUOK, FBXL4, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP		
Mitochondriale Myopathie / Enzephalopathie [CO]	3 - 5 Wo	E
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		
Mitochondriengenom [CO]	3 - 5 Wo	E
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		
Nukleär-kodierte mitochondriale Erkrankungen *	4 - 6 Wo	E
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		
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		
Pyruvatdehydrogenase-Mangel (PDHD) *	3 - 5 Wo	E
Gen-Panel: ID393.00, 7 Gene (10,0 kb)		
DLAT, DLD, LIAS, PDHA1, PDHB, PDHX, PDP1		

Erkrankung/Diagnostik	Dauer	Material
Multisystem-Fehlbildungssyndrome		
Alagille-Syndrom (ALGS) *	3 - 5 Wo	E
Gen-Panel: ID112.00, 2 Gene (11,1 kb)		
JAG1, NOTCH2		
Alport-Syndrom (ATS) *	3 - 5 Wo	E
Gen-Panel: ID099.00, 4 Gene (21,0 kb)		
COL4A3, COL4A4, COL4A5, MYH9		
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		
CHARGE-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID307.00, 3 Gene (12,9 kb)		
CHD7, SEMA3E, TBX22		
Coffin-Siris-Syndrom (CSS) *	3 - 5 Wo	E
Gen-Panel: ID118.02, 14 Gene (46,5 kb)		
ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11		
Cornelia-de-Lange-Syndrom (CDLS) *	3 - 5 Wo	E
Gen-Panel: ID033.02, 8 Gene (32,0 kb)		
ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS		
Fehlbildungssyndrome mit überwiegend fazialer Beteiligung *	3 - 5 Wo	E
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		
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 Defekt der oxidativen Phosphorylierung (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 Glykosylierungsstörung (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: ID265.00, 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Rubinstein-Taybi-Syndrom (RSTS) *	3 - 5 Wo	E
Gen-Panel: ID142.01, 3 Gene (24,3 kb)		
CREBBP, EP300, SRCAP		

Erkrankung/Diagnostik	Dauer	Material
Multisystem-Fehlbildungssyndrome		
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
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
Tuberöse Sklerose (TSC) * Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
VACTERL-Assoziation * 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	3 - 5 Wo	E
Weill-Marchesani-Syndrom (WMS) * Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
Zellweger-Syndrom (ZWS) * Gen-Panel: ID084.00, 14 Gene (22,3 kb) ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	3 - 5 Wo	E
Muskelerkrankungen		
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) 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	4 - 6 Wo	E
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) 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 Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46 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	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN) *	4 - 6 Wo	E
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		
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		
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN) *	3 - 5 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)		
EGR2, 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 hereditäre motorische Neuronopathie (HMND, HMNR) *	3 - 5 Wo	E
Gen-Panel: ID254.01		
Distale hereditäre motorische Neuronopathie (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		
Distale motorische Neuronopathie, autosomal-dominant (HMND): 15 Gene (38,4 kb)		
BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1		
Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 10 Gene (14,4 kb)		
ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, 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		
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		

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
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		
Central-Core-Myopathie (CCD): 1 Gen (15,1 kb)		
RYR1		
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		
Letales kongenitales Kontraktursyndrom (LCCS) *	3 - 5 Wo	E
Gen-Panel: ID197.00, 12 Gene (34,6 kb)		
ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42		
Muskeldystrophie-Dystroglykanopathie (MDDG) *	3 - 5 Wo	E
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		
Muskeldystrophie, Typ Duchenne oder Becker (DMD, BMD) *	2 - 4 Wo	E
Gen-Panel: ID256.00, 1 Gen (11,1 kb)		
DMD		
Muskelerkrankungen, umfassende Diagnostik *	4 - 6 Wo	E
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		

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
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
Spinale Muskelatrophie (SMA) * Gen-Panel: ID152.02 Spinale Muskelatrophie (SMA): 39 Gene (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, 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): 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	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
Neurodegenerative Erkrankungen		
Alzheimer-Krankheit (AD) * Gen-Panel: ID157.01, 7 Gene (16,8 kb) ABCA7, ADAM10, APOE, APP, PSEN1, PSEN2, TF	3 - 5 Wo	E
Amyotrophe Lateralsklerose (ALS) * Gen-Panel: ID209.03, 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	3 - 5 Wo	E
Basalganglien-Kalzifikation (IBGC) * Gen-Panel: ID327.00, 6 Gene (11,2 kb) JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1	3 - 5 Wo	E
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 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	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN) *	3 - 5 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)		
EGR2, MPZ, CNTNAP1		
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik *	4 - 6 Wo	E
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)		
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		
Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6, HMSN6): 3 Gene (4,5 kb)		
MFN2, PDXK, SLC25A46		
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		
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 hereditäre motorische Neuronopathie (HMND, HMNR) *	3 - 5 Wo	E
Gen-Panel: ID254.01		
Distale hereditäre motorische Neuronopathie (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		
Distale motorische Neuronopathie, autosomal-dominant (HMND): 15 Gene (38,4 kb)		
BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1		
Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 10 Gene (14,4 kb)		
ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1		
Dystonie (DYT) *	3 - 5 Wo	E
Gen-Panel: ID128.04		
Dystonie (DYT): 25 Gene (55,9 kb)		
ACTB, ADCY5, ANO3, AOPEP, ATP1A3, COL6A3, ECHS1, GCH1, GNAL, HPCA, KCTD17, KMT2B, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VPS16		
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		

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Episodisches Schmerzsyndrom (FEPS) *	3 - 5 Wo	E
Gen-Panel: ID268.00, 4 Gene (20,5 kb)		
SCN9A, SCN10A, SCN11A, TRPA1		
Essentieller Tremor (ETM) *	3 - 5 Wo	E
Gen-Panel: ID195.01, 4 Gene (16,6 kb)		
DRD3, FUS, SCN4A, TENM4		
Frontotemporale Demenz (FTD) *	3 - 5 Wo	E
Gen-Panel: ID310.01, 19 Gene (29,3 kb)		
C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP		
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		
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		
Hereditäre Neuropathien, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID374.00		
Hereditäre Neuropathien, umfassende Diagnostik: 134 Gene (343,6 kb)		
AARS1, ABCA1, ACOX1, AIFM1, AR, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, 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, 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): 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		
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		

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Hereditäre sensorische und autonome Neuropathie (HSAN, HSN) *	3 - 5 Wo	E
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		
Hirnatrophie und demyelinisierende Erkrankungen des Gehirns *	3 - 5 Wo	E
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		
Hyperekplexie (HKPX) *	3 - 5 Wo	E
Gen-Panel: ID216.00, 9 Gene (16,0 kb)		
ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1		
Hypomyelinisierende Leukodystrophie (HLD) *	3 - 5 Wo	E
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		
Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik *	4 - 6 Wo	E
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		
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		
Neurologische Entwicklungsstörungen (NED), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID358.00, 169 Gene (431,8 kb)		
ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDFT1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNPK, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, 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, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6		
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Parkinson-Krankheit (PARK) *	3 - 5 Wo	E
Gen-Panel: ID077.02		
Parkinson-Krankheit (PARK): 37 Gene (93,8 kb)		
ADH1C, ATP13A2, ATP1A3, ATP6AP2, CHCHD2, DCTN1, DNAJC6, EIF4G1, FBXO7, GBA, GCH1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, POLG, PRKRA, PRKN, PSAP, RAB39B, SLC18A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SYNJ1, TAF1, TH, TRPM7, UCHL1, UQCRC1, VPS13C, VPS35		
Parkinson-Krankheit (PARK), early-onset: 9 Gene (30,3 kb)		
ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, SYNJ1, VPS13C		
Parkinson-Krankheit (PARK), late-onset: 12 Gene (27,3 kb)		
ADH1C, CHCHD2, EIF4G1, GBA, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, SNCA, UCHL1, VPS35		
Dystonie-Parkinsonismus (DYT): 9 Gene (18,3 kb)		
ATP1A3, GCH1, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH		
Schlafstörungen *	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: ID148.04		
Spastische Paraplegie (HSP, SPG): 67 Gene (138,1 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, 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: 50 Gene (101,2 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, 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: ID152.02		
Spinale Muskelatrophie (SMA): 39 Gene (87,9 kb)		
AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, 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): 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		
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Alport-Syndrom (ATS) *	3 - 5 Wo	E
Gen-Panel: ID099.00, 4 Gene (21,0 kb)		
COL4A3, COL4A4, COL4A5, MYH9		
Aminoazidurie *	3 - 5 Wo	E
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		
Atypisches hämolytisch-urämisches Syndrom (AHUS) *	3 - 5 Wo	E
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		
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		
Bartter-Syndrom (BARTS) *	3 - 5 Wo	E
Gen-Panel: ID156.01, 8 Gene (17,7 kb)		
BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3		
Branchiootorenales Syndrom (BOR) *	3 - 5 Wo	E
Gen-Panel: ID315.00, 5 Gene (10,2 kb)		
EYA1, SALL1, SIX1, SIX5, TFAP2A		
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen *	3 - 5 Wo	E
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		
Diabetes insipidus *	3 - 5 Wo	E
Gen-Panel: ID322.00, 5 Gene (8,4 kb)		
AQP2, AVP, AVPR2, SLC12A1, WFS1		
Endokrine Hypertonie *	3 - 5 Wo	E
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		
Fraser-Syndrom (FRASRS) *	3 - 5 Wo	E
Gen-Panel: ID317.00, 3 Gene (24,8 kb)		
FRAS1, FREM2, GRIP1		
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		
Glomerulonephritis *	3 - 5 Wo	E
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
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
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) 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	3 - 5 Wo	E
Joubert-Syndrom (JBTS) * 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	4 - 6 Wo	E
Kongenitale Anomalien der Niere und ableitenden Harnwege (CAKUT) * 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	4 - 6 Wo	E
Meckel-Syndrom (MKS) * Gen-Panel: ID032.02, 13 Gene (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 Wo	E
Metabolische Nierenerkrankungen * Gen-Panel: ID705.00, 29 Gene (53,4 kb) AGXT, APOA1, ATP7B, B2M, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, FAH, FGA, GALT, GLA, GRHPR, GSN, HOGA1, KCNJ1, LYZ, MEFV, MVK, NLRC4, NLRP12, NLRP3, OCRL, PLCG2, SLC12A1, SLC26A1, TNFRSF1A, TTR	3 - 5 Wo	E
Mikrohämaturie * Gen-Panel: ID385.00, 11 Gene (44,0 kb) CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FN1, INF2, MYH9, PIGA, PIGT, UMOD	3 - 5 Wo	E
Nephroblastom und Wilms-Tumor (WT) * 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	3 - 5 Wo	E
Nephrokalzinose * Gen-Panel: ID361.00, 29 Gene (54,1 kb) ADCY10, AGXT, ALPL, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, FAH, FAM20A, GRHPR, HOGA1, KCNJ1, MAGED2, OCRL, OXGR1, RRAGD, SLC12A1, SLC4A1, SLC34A1, SLC34A3, VIPAS39, VPS33B	3 - 5 Wo	E
Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS) * 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	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Nephronophthise (NPHP) * 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	3 - 5 Wo	E
Nierenzellkarzinom * 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	3 - 5 Wo	E
Polyzystische Lebererkrankung (PCLD) * 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	3 - 5 Wo	E
Polyzystische Nierenerkrankung (PKD) * 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	3 - 5 Wo	E
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) * 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	3 - 5 Wo	E
Renale Ziliopathien, umfassende Diagnostik * 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	4 - 6 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 Periodisches Fiebersyndrom mit Amyloidose: 3 Gene (6,8 kb) MEFV, NLRP3, TNFRSF1A	3 - 5 Wo	E
Renale Hypodysplasie, Aplasie und Agenesie * 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	3 - 5 Wo	E
Renale tubuläre Azidose (RTA) * Gen-Panel: ID297.00, 9 Gene (18,5 kb) ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Renale tubuläre Dysgenesie (RTD) *	2 - 4 Wo	E
Gen-Panel: ID316.00, 4 Gene (7,7 kb)		
ACE, AGT, AGTR1, REN		
Renotubuläres Fanconi-Syndrom (FRTS) *	3 - 5 Wo	E
Gen-Panel: ID359.00, 7 Gene (10,4 kb)		
CTNS, EHHADH, GATM, HNF4A, NDUFAF6, SLC2A2, SLC34A1		
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		
Thrombotische Mikroangiopathie (TMA) *	3 - 5 Wo	E
Gen-Panel: ID707.00		
Thrombotische Mikroangiopathie (TMA): 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		
Thrombotisch-thrombozytopenische Purpura (TTP): 1 Gen (4,3 kb)		
ADAMST13		
Atypisches hämolytisch-urämisches Syndrom (AHUS): 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		
Tubulointerstitielle Nierenerkrankung, autosomal-dominant (ADTKD) *	3 - 5 Wo	E
Gen-Panel: ID296.00, 6 Gene (8,8 kb)		
DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD		
Urolithiasis, Nephrolithiasis und Nephrokalzinose *	3 - 5 Wo	E
Gen-Panel ID231.04		
Urolithiasis, Nephrolithiasis und Nephrokalzinose: 37 Gene (68,8 kb)		
ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, BSND, CASR, CLCN5, CLDN16, CLDN19, CLCNKB, CYP24A1, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, OCRL, OXGR1, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, XDH		
Hyperkalziurie: 15 Gene (30,2 kb)		
ADCY10, BSND, CASR, CLCN5, 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		
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		
Vesikoureteraler Reflux (VUR) *	3 - 5 Wo	E
Gen-Panel: ID314.00, 10 Gene (33,7 kb)		
DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB		
Zystische Nierenerkrankungen, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID100.09		
Zystische Nierenerkrankungen, umfassende Diagnostik: 53 Gene (165,3 kb)		
ALG5, ALG6, ALG8, ALG9, ANKS6, CEP83, CEP164, CEP290, COL4A1, CRB2, CYS1, 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, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, XPNPEP3, ZNF423		
Polyzystische Nierenerkrankung (PKD): 9 Gene (41,8 kb)		
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		
Medulläre zystische Nierenerkrankung (MCKD, 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		

Erkrankung/Diagnostik	Dauer	Material
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		
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)	4 - 6 Wo	E
BHLHA9, FBLN1, GJA1, GLI3, HOXD13, LMBR1, LRP4, NECTIN4		
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		

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
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
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) 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	3 - 5 Wo	E
Klippel-Feil-Syndrom (KFS) * Gen-Panel: ID207.00, 5 Gene (12,5 kb) GDF3, GDF6, MEOX1, MYO18B, PAX1	3 - 5 Wo	E
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 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	3 - 5 Wo	E
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 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	3 - 5 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
Lippen-, Kiefer- und Gaumenspalte (OFC, CLP) * 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	4 - 6 Wo	E
Mandibulofaziale Dysostose (MFD) * 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	3 - 5 Wo	E
Multiple epiphysäre Dysplasie (EDM) * Gen-Panel: ID202.02, 11 Gene (27,7 kb) CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CSGALNACT1, EIF2AK3, KIF7, MATN3, SLC26A2	3 - 5 Wo	E
Multiples Pterygium-Syndrom * Gen-Panel: ID158.01, 8 Gene (16,8 kb) CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Orofaziodigitales Syndrom (OFD) * Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 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
Osteopetrose (OPT) und ähnliche Knochenerkrankungen * 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	3 - 5 Wo	E
Osteoporose * Gen-Panel: ID115.02, 15 Gene (39,0 kb) CALCR, COPB2, COL1A1, COL1A2, ESR1, LGR4, LRP5, NOTCH2, PLS3, NHERF1, SGMS2, SLC34A1, VDR, WNT1, WNT11	3 - 5 Wo	E
Pierre-Robin-Sequenz * Gen-Panel: ID294.00, 33 Gene (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E
Polydaktylie, nicht-syndromale Form * Gen-Panel: ID166.02, 9 Gene (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141	3 - 5 Wo	E
Rubinstein-Taybi-Syndrom (RSTS) * Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
Skelettdysplasie, schwere Form * 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, 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	4 - 6 Wo	E
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

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD) *	3 - 5 Wo	E
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		
Spondylokostale Dysostose (SCDO) *	3 - 5 Wo	E
Gen-Panel: ID227.00, 7 Gene (14,4 kb)		
DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6		
Weill-Marchesani-Syndrom (WMS) *	3 - 5 Wo	E
Gen-Panel: ID230.00, 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
3M-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID214.00, 3 Gene (12,4 kb)		
CCDC8, CUL7, OBSL1		
Stoffwechselerkrankungen		
Aminoazidurie *	3 - 5 Wo	E
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		
Amyloidose *	3 - 5 Wo	E
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		
Coenzym-Q10-Mangel (COQ10D) *	3 - 5 Wo	E
Gen-Panel: ID225.01, 15 Gene (18,7 kb)		
ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2		
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen *	3 - 5 Wo	E
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		
Folatstoffwechselstörung *	3 - 5 Wo	E
Gen-Panel: ID334.00, 10 Gene (18,7 kb)		
CBS*, FOLR1, FOLR2, FTCD, MTHFD1, MTHFR, MTR, MTRR, SLC19A1, SLC46A1		
Glykogenspeicherkrankheit (GSD) *	3 - 5 Wo	E
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		
Glykosylphosphatidylinositol (GPI)-Biosynthesedefekt (GPIBD) *	3 - 5 Wo	E
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Homocystinurie * Gen-Panel: ID191.01, 9 Gene (15,3 kb) ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1	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
Hyperkalzämie * Gen-Panel: ID262.00, 8 Gene (14,6 kb) AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1	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
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) * Gen-Panel: ID292.00, 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung * 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	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
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) 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	3 - 5 Wo	E
Kombinierter Defekt der oxidativen Phosphorylierung (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
Kongenitale Glykosylierungsstörung (CDG) * Gen-Panel: ID035.03 Kongenitale Glykosylierungsstörung (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	3 - 5 Wo	E
MODY-Diabetes * Gen-Panel: ID048.01, 14 Gene (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 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

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Muskeldystrophie-Dystroglykanopathie (MDDG) *	3 - 5 Wo	E
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		
Metabolische Epilepsien *	4 - 6 Wo	E
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		
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		
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		
Porphyrie *	3 - 5 Wo	E
Gen-Panel: ID153.01, 10 Gene (12,8 kb)		
ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS		
Pulmonale Surfactant-Stoffwechselstörung (SMDP) *	3 - 5 Wo	E
Gen-Panel: ID168.01, 6 Gene (12,0 kb)		
ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB		
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		
Periodisches Fiebersyndrom mit Amyloidose: 3 Gene (6,8 kb)		
MEFV, NLRP3, TNFRSF1A		
Speicherkrankheiten mit Herzbeteiligung *	3 - 5 Wo	E
Gen-Panel: ID149.02		
Speicherkrankheiten mit Herzbeteiligung: 17 Gene (25,1 kb)		
APOA1, ATP7B, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR		
Amyloidose (AMYLD): 6 Gene (6,4 kb)		
APOA1, B2M, FGA, GSN, LYZ, TTR		
Hämochromatose (HFE): 6 Gene (7,3 kb)		
FTH1, HAMP, HFE, HJV, SLC40A1, TFR2		
Kardiale Glykogenspeicherkrankheit (GSD): 3 Gene (5,8 kb)		
GAA, LAMP2, PRKAG2		

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
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		
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		
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		
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Erbliche Tumorerkrankungen, umfassende Diagnostik * Gen-Panel: ID018.04, 353 Gene (779,1 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, 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, GPC3, GPC4, 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, RPS17, 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, USP45, UVSSA, VHL, VPS45, WAS, WRAP53, WRN, WT1, WWP1, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, ZBTB24, ZCCHC8, ZNF687, ZSWIM7	4 - 6 Wo	E
Gastrointestinaler Stromatumor (GIST) * Gen-Panel: ID226.00, 8 Gene (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 Wo	E
Glioblastom (GLM) * 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	3 - 5 Wo	E
Gorlin-Syndrom (BCNS) und ähnliche Krankheitsbilder * Gen-Panel: ID174.02, 8 Gene (25,8 kb) BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 Wo	E
Hypophysenadenom (PITA) * Gen-Panel: ID387.01, 21 Gene (47,3 kb) AIP, CDH23, CDKN1B, DICER1, EPCAM, GNAS, GPR101, MAX, MLH1, MSH2, MSH6, MEN1, PMS2, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, USP8	3 - 5 Wo	E
Kolorektales Karzinom und Polyposis* Gen-Panel ID006.09 Kolorektales Karzinom und Polyposis: 23 Gene (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Kolorektales Karzinom: 18 Gene (55,7 kb) APC, ATM, CHEK2, BMPR1A, EPCAM, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11 Kolorektale Polyposis: 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
Kolorektales Karzinom mit Mikrosatelliteninstabilität (MSI) * Gen-Panel: ID283.00, 9 Gene (25,4 kb) EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE	3 - 5 Wo	E
Krebserkrankungen im Kindesalter * 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	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Konstitutionelles MMR-Defizienz-Syndrom (CMMRDS, MMRCS) * Gen-Panel: ID362.00, 5 Gene (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	2 - 4 Wo	E
Kutanes malignes Melanom (CMM) * Gen-Panel: ID193.01, 12 Gene (26,4 kb) BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3	3 - 5 Wo	E
Lynch-Syndrom (LYNCH, HNPCC) * Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	3 - 5 Wo	E
Lungenkarzinom * 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	4 - 6 Wo	E
Magenkarzinom * 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	3 - 5 Wo	E
Mammakarzinom * Gen-Panel: ID021.02, 12 Gene (41,0 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53	3 - 5 Wo	E
Mamma- und Ovarialkarzinom (HBOC) * Gen-Panel: ID003.04 Mamma- und Ovarialkarzinom (HBOC): 19 Gene (62,5 kb) ATM, BARD1, BRIP1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53 Mammakarzinom: 10 Gene (38,9 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, STK11, TP53 Ovarialkarzinom: 14 Gene (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 Wo	E
Medulloblastom (MDB) * 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	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
Nephroblastom und Wilms-Tumor (WT) * 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	3 - 5 Wo	E
Neuroendokrine Neoplasie * 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	3 - 5 Wo	E
Neurofibromatose (NF) * Gen-Panel: ID210.00, 3 Gene (11,6 kb) NF1, NF2, SPRED1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Nierenzellkarzinom * 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	3 - 5 Wo	E
Ovarialkarzinom * Gen-Panel: ID004.04, 14 Gene (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 Wo	E
Pankreaskarzinom * 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	3 - 5 Wo	E
Phäochromozytom-Paragangliom-Syndrom (PPGL) * Gen-Panel: ID042.03, 14 Gene (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E
Plasmozytom * 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	3 - 5 Wo	E
Polyposis-Syndrom (PS, FAP) * Gen-Panel: ID005.06, 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
Prostatakarzinom * 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	3 - 5 Wo	E
Sarkom der Weichteile und des Skelettsystems * 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	4 - 6 Wo	E
Schilddrüsenkarzinom * 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	3 - 5 Wo	E
Tuberöse Sklerose (TSC) * Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
Urothelkarzinom * 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	3 - 5 Wo	E
Xeroderma pigmentosum (XP) * Gen-Panel: ID282.00, 10 Gene (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 Wo	E
Ziliopathien		
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
Joubert-Syndrom (JBTS) * 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	4 - 6 Wo	E
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 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	3 - 5 Wo	E
Meckel-Syndrom (MKS) * Gen-Panel: ID032.02, 13 Gene (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 Wo	E
Nephronophthise (NPHP) * 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	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Ziliopathien		
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) * 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	4 - 6 Wo	E
Senior-Loken-Syndrom (SLSN) * Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E
Renale Ziliopathien, umfassende Diagnostik * 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	4 - 6 Wo	E
Präventionsdiagnostik: Carrier-Screening		
Carrier-Screening * Gen-Panel: ID350.00, 625 Gene (1591,4 kb) AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACOX1, ACSF3, ADA, ADAMTS13, ADAMTS2, ADGRG1, ADGRV1, AFF2, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMT, ANK1, ANO10, ANTXR2, 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, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRIP1, BSND, BTD, BTK, CA2, CAPN3, CASR, CBS, CC2D2A, CCDC88C, CD3D, CD3E, CD40LG, CDH23, CEP290, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNG, CIITA, CLCN1, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL1A2, COL27A1, COL4A3, COL4A4, COL4A5, COL7A1, COQ2, COQ8A, CPS1, CPT1A, CPT2, CRB1, CRLF1, CRTAP, CSTB, CTNS, CTSC, CTSD, CTSK, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP27A1, CYP27B1, D2HGDH, DBT, DCLRE1C, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DHDDS, DKC1, DLD, DLL3, DMD, DMP1, DNAH5, DNAI1, DNAI2, DNAJC19, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPYD, DSP, DUOX2, DUOXA2, DYNC2H1, DYSF, EDA, 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, F8, F9, FAH, FAM126A, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCG, FBLN5, FBP1, FGA, FGB, FGD4, FGG, FH, FKRP, FKTN, FLNA, FMO3, FOLR1, FOXN1, FOXP3, FRAS1, FREM2, FUCA1, FXN, G6PC1, G6PC3, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDF5, GFM1, GJA1, GJB1, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNE, GNPTAB, GNPTG, GNS, GP1BA, GP9, GRHPR, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HESX1, HEXA, HEXB, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HOGA1, HPD, HPRT1, HPS1, HPS3, HPS4, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPG2, HYAL1, HYLS1, IDS, IDUA, IGBP1, IGF1, IGHMBP2, IGSF1, IL2RG, IL7R, INPP5E, INS, INSR, INVS, IQCB1, ITGA6, ITGB4, IVD, IYD, JAG1, JAK3, KCNJ1, KCNJ11, KCNQ1, KCNQ2, KCTD7, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LARGE1, LBR, LCA5, LDLR, LDLRAP1, LHCGR, LHX3, LIFR, LIPA, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MED12, MED17, MEFV, MESP2, MFSD8, MGAT2, MID1, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPI, MPL, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTTP, MUTYH, MVK, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFAF5, NDUFS6, NEU1, NEUROG3, NHLRC1, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR2E3, NTRK1, NUP62, OAT, OCA2, OCRL, OFD1, OPA3, OSTM1, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRT2, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRT2, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A1, SLC4A11, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMARCAL1, SMPD1, SNAP29, SP110, SPR, SRD5A2, ST3GAL5, STAR, STRA6, SUCLG1, SUMF1, SUOX, TAFAZZIN, TAT, TBCE, TCIRG1, TECPR2, TF, TFR2, TG, TGM1, TH, TIMM8A, TK2, TMEM216, TMEM67, TNFRSF11B, TNNT1, TPO, TPP1, TREX1, TRHR, TRIM32, TRIM37, TRMU, TSEN54, TSHB, TSHR, TSPYL1, TTC37, TTN, TTPA, TWNK, TYMP, TYR, TYRP1, UBA1, UBR1, UGT1A1, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WAS, WNT10A, WNT3, WNT7A, WRN, WT1, XPA, XPC, ZIC3, ZMPSTE24, ZNF469	4 - 6 Wo	E

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, MSTO1, 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, 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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
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		