SYNLAB Humangenetik

Stand: 12/2025

Next Generation Sequencing (NGS)

Panel-Verzeichnis

Whole Exome Sequencing (WES)

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 *	3 - 5 Wo	E
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		
Katarakt (CTRCT) *	3 - 5 Wo	E
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		
Kongenitale extraokuläre Muskelfibrose (CFEOM) *	3 - 5 Wo	E
Gen-Panel: ID063.00, 5 Gene (10,5 kb)		
COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3		
Kongenitaler Nystagmus (NYS) *	3 - 5 Wo	E
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		
Kongenitale stationäre Nachtblindheit (CSNB) *	3 - 5 Wo	E
Gen-Panel: ID267.00, 14 Gene (39,2 kb)		
CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, SAG, RHO, SLC24A1, TRPM1		
Lebersche hereditäre Optikusneuropathie (LHON) #	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		
Lebersche kongenitale Amaurose (LCA) *	3 - 5 Wo	E
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		
Makuladystrophie (MD) *	3 - 5 Wo	E
Gen-Panel: ID139.03, 22 Gene (49,6 kb)		
ABCA4, BEST1, CDH3, CDHR1, CHST6, CLEC3B, CNGB3, CRB1, CRX, CTNNA1, ELOVL4, IMPG1, IMPG2, MAPKAPK3, MFSD8, PROM1, PRPH2, RDH8, RDH12, RP1L1, SIX6, TIMP3		
Morbus Stargardt (STGD) *	3 - 5 Wo	E
Gen-Panel: ID102.01, 4 Gene (11,4 kb)		
ABCA4, ELOVL4, PROM1, PRPH2		
Optikusatrophie (OPA) *	3 - 5 Wo	E
Gen-Panel ID081.05		
Optikusatrophie (OPA): 38 Gene (60,6 kb)		
ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, KLC2, MCAT, MECR, MFF, MFN2, MIEF1, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, RTN4IP1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, SSBP1, TBCE, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1, ZNHIT3		
Optikusatrophie (OPA) nicht-syndromal: 12 Gene (18,5 kb)		
ACO2, AFG3L2, DNM1L, MCAT, MECR, MIEF1, OPA1, OPA3, RTN4IP1, SSBP1, TMEM126A, YME1L1		
Syndrome mit Optikusatrophie: 29 Gene (46,6 kb)		
ATP1A3, C19ORF12, CISD2, DNAJC30, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, TBCE, TIMM8A, UCHL1, WFS1, ZNHIT3		

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
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
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
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 (21,1 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
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		
Vitreoretinopathie *	3 - 5 Wo	E
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		
Walker-Warburg-Syndrom (WWS, MDDGA) *	3 - 5 Wo	E
Gen-Panel: ID178.00, 14 Gene (23,7 kb)		
B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1		
Weill-Marchesani-Syndrom (WMS) *	3 - 5 Wo	E
Gen-Panel: ID230.00, 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD) *	3 - 5 Wo	E
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		
Bindegewebserkrankungen		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID137.06		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik: 85 Gene (283,3 kb)		
ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ASPH, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, DCC, DLG4, DSE, EFEMP1, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, IPO8, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NKAP, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THBS2, THSD4, TNXB, ZNF469		
Marfan-Syndrom (MFS): 3 Gene (11,8 kb)		
FBN1, TGFBR2, TGFBR1		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 18 Gene (45,7 kb)		
ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Cutis laxa-Syndrom (ARCL, ADCL): 12 Gene (29,4 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
Stickler-Syndrom (STL): 6 Gene (22,0 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2		
Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Cutis laxa (ARCL, ADCL) *	3 - 5 Wo	E
Gen-Panel: ID109.03, 13 Gene (32,1 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1, RIN2		
Ehlers-Danlos-Syndrom (EDS) *	3 - 5 Wo	E
Gen-Panel: ID039.06		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 9 Gene (39,6 kb)		
C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1, THBS2		
Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469		
Hereditäre hämorrhagische Teleangiektasie (HHT) *	3 - 5 Wo	E
Gen-Panel: ID155.01, 7 Gene (15,6 kb)		
ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4		

Erkrankung/Diagnostik	Dauer	Material
Bindegewebserkrankungen		
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.08 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 40 Gene (117,6 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (12,6 kb) BGN, IPO8, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit thorakalem Aortenaneurysma: 31 Gene (96,5 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, IPO8, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E
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 ID194.06 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 50 Gene (157,9 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP1, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 17 Gene (42,1 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Stickler-Syndrom (STL): 6 Gene (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Syndrome mit marfanoidem Habitus: 20 Gene (52,2 kb) CBS, DLG4, EFEMP1, EFEMP2, FBN1, FBN2, NKAP, NPR2, MED12, PLOD1, PRDM5, SKI, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZDHHC9	4 - 6 Wo	E
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 (21,1 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) * Gen-Panel: ID020.02 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 17 Gene (44,1 kb) ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	3 - 5 Wo	E
Endokrine Störungen		
Adipositas * Gen-Panel: ID183.03 Adipositas: 57 Gene (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 18 Gene (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 40 Gene (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
Adrenogenitales Syndrom (AGS, CAH) * Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
Diabetes insipidus * Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E
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

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
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
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

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
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
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
Variante der Geschlechtsentwicklung (DSD) * Gen-Panel ID117.04 Variante der Geschlechtsentwicklung (DSD): 55 Gene (106,2 kb) AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR3C1, NR5A1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX3, SOX8, SOX9, SOX10, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 20 Gene (36,1 kb) ANOS1, AKR1C2, AKR1C4, AR, CBX2, DHH, DHX37, DMRT1, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 7 Gene (8,7 kb) NR2F2, NR5A1, SOX3, SOX9, SRY, WT1, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 41 Gene (80,2 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, NR3C1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX9, SOX10, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
Wachstumshormonmangel (IGHD, CPHD) * Gen-Panel: ID211.02 Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNPC3, OTX2, POU1F1, PROP1, SEMA3A, SOX3 Isolierter Wachstumshormonmangel (IGHD): 5 Gene (6,4 kb) BTK, GH1, GHRHR, GHSR, SOX3 Kombinierter Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Adipositas * Gen-Panel: ID183.03 Adipositas: 57 Gene (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 18 Gene (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 40 Gene (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
Autismus-Spektrum-Störungen * Gen-Panel: ID076.04, 168 Gene (664,6 kb) AHDC1, ADNP, ADSL, AFF2, AGO1, ALDH5A1, ANK2, ANKRD11, ARHGEF9, ARID1B, ARX, ASH1L, ASTN2, ASXL3, ATP1A1, AUTS2, BAZ2B, BCKDK, BCL11A, BRSK2, CACNA1C, CAPRIN1, CASK, CASZ1, CC2D1A, CDKL5, CELF4, CHD2, CHD7, CHD8, CIC, CNOT3, CNTN4, CNTNAP2, CREBBP, CSDE1, CSNK2A1, CTNNB1, CUL3, DDX3X, DEAF1, DHCR7, DIP2A, DLGAP2, DNMT3A, DPP6, DSCAM, DYNC1H1, DYRK1A, EBF3, EHMT1, EIF4E, EP300, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, GIGYF1, GLRA2, GRIA2, GRIN2A, GRIN2B, HERC2, HNRNPU, IL1RAPL1, IQSEC2, IRF2BPL, KATNAL2, KCNQ3, KDM5A, KDM5C, KDM6B, KMT2A, KMT2C, KMT2E, KMT5B, MAGEL2, MAOA, MBD5, MBOAT7, MECP2, MED13, MED13L, MEF2C, MEIS2, MYT1L, NAA15, NEXMIF, NF1, NHS, NIPBL, NLGN1, NLGN3, NLGN4X, NOVA2, NR1I3, NR4A2, NRXN1, NRXN2, NRXN3, NSD1, OPHN1, PAH, PAX5, PCDH19, PDZD8, PHF12, PHF2, PHF21A, PHF3, POGZ, PPP2R5D, PSMD12, PTCHD1, PTEN, RAB39B, RAI1, RELN, RERE, RFX3, RIMS1, RORB, RPL10, SATB2, SCN1A, SCN2A, SEMA5A, SETD2, SETD5, SGSH, SHANK2, SHANK3, SLC6A1, SLC6A8, SLC9A6, SLC9A9, SMARCB1, SMARCC2, SON, SOX5, SPAST, STXBP1, SYN1, SYNGAP1, TANC2, TBL1XR1, TBR1, TCF20, TCF4, TLK2, TMLHE, TRIP12, TRPC6, TRRAP, TSC1, TSC2, UBE3A, UPF3B, VAMP2, VPS13B, WAC, WDFY3, ZMYM3, ZMYND8, ZNF292, ZNF462, ZSWIM6	4 - 6 Wo	E
Coffin-Siris-Syndrom (CSS) * Gen-Panel: ID118.01, 14 Gene (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 Wo	E
Cornelia-de-Lange-Syndrom (CDLS) * Gen-Panel: ID033.02, 8 Gene (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 Wo	E
FG-Syndrom (FGS) * Gen-Panel: ID215.00, 3 Gene (17,2 kb) CASK, FLNA, MED12	3 - 5 Wo	E
Großwuchs, umfassende Diagnostik * Gen-Panel: ID299.01, 64 Gene (211,5 kb) ABCC9, AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, FBN1, FIBP, GFAP, GLI3, GPC3, GPC4, GRIA3, H1-4, HEPACAM, HERC1, HUWE1, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIGA, PIK3CA, PIK3R2, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SHANK, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMEM94, TRIP12, UPF3B, ZBTB20, ZDHHC9	4 - 6 Wo	E
Großwuchssyndrome (SOTOS, BWS) * Gen-Panel: ID073.05 Großwuchssyndrome (SOTOS, BWS): 15 Gene (41,2 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, PDGFRB, PTEN, SETD2, SUZ12, RNF125 Sotos-ähnliche Großwuchssyndrome: 6 Gene (23,1 kb) EED, EZH2, NFIX, NSD1, SETD2, SUZ12 Beckwith-Wiedemann-ähnliche Großwuchssyndrome: 5 Gene (10,1 kb) CDKN1C, DIS3L2, GPC3, GPC4, OFD1	3 - 5 Wo	E
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) * Gen-Panel: ID292.00, 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD, IDD) * 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	4 - 6 Wo	E

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, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NEMF, NSUN2, NSUN6, NUDT2, OTUD6B, PDE2A, PDZD8, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, PUS7, RSRC1, RUSC2, SCAPER, SLC45A1, SLC6A17, ST3GAL3, TAF13, TAF2, TECR, TMEM94, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, WIPI2, ZBTB11, ZC3H14		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv, nicht-syndromal (MRT): 58 Gene (157,9 kb)		
ADAT3, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31, FERRY3, FMN2, GRIA1, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NSUN2, NSUN6, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, ZBTB11, ZC3H14		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv, syndromal (IDD): 20 Gene (48,5 kb)		
ABCA2, ALG14, CPE, DEAF1, FBXL3, GNB5, IQSEC1, LGI3, NEMF, NUDT2, OTUD6B, PDE2A, PDZD8, PUS7, SCAPER, SLC45A1, TAF2, TMEM94, WIPI2, ZBTB11		
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS) *	4 - 6 Wo	E
Gen-Panel: ID038.06		
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS): 77 Gene (194,6 kb)		
ACSL4, AFF2, AP1S2, ARX, ATP6AP2, ATRX, BRWD3, CASK, CLCN4, CLIC2, CNKSR2, CSTF2, CUL4B, DDX3X, DLG3, EIF2S3, FAM50A, FGD1, FGF13, FMR1, FRMPD4, FTSJ1, GDI1, GLRA2, GRIA3, HCFC1, HNRNPH2, HS6ST2, HUWE1, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIF4A, KLHL15, LAS1L, MECP2, MED12, MID2, MSL3, NEXMIF, NKAP, NONO, OGT, OPHN1, PAK3, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, RPS6KA3, SLC16A2, SLC9A6, SLC9A7, SLITRK2, SMS, STEEP1, SYN1, SYP, TAF1, THOC2, TSPAN7, UBE2A, UPF3B, USP9X, USP27X, WNK3, ZMYM3, ZC4H2, ZDHHC9, ZFX, ZNF711		
Intellektuelle Entwicklungsstörung, X-chromosomal, nicht-syndromal (XLID, MRX): 29 Gene (77,8 kb)		
ACSL4, AFF2, ARX, BRWD3, DLG3, FGF13, FRMPD4, FTSJ1, GDI1, HCFC1, IL1RAPL1, IQSEC2, KIF4A, KLHL15, MID2, NEXMIF, OGT, PAK3, RAB39B, RPS6KA3, SLC9A7, STEEP1, SYN1, SYP, THOC2, TSPAN7, USP9X, USP27X, ZNF711		
Intellektuelle Entwicklungsstörung, X-chromosomal, syndromal (MRXS): 52 Gene (130,8 kb)		
AFF2, AP1S2, ARX, ATP6AP2, ATRX, CASK, CLCN4, CLIC2, CNKSR2, CSTF2, CUL4B, DDX3X, EIF2S3, FAM50A, FGD1, FMR1, GLRA2, GRIA3, HNRNPH2, HS6ST2, HUWE1, IGBP1, KDM5C, LAS1L, MECP2, MED12, MSL3, NKAP, NONO, OPHN1, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, SLC16A2, SLC9A6, SLITRK2, SMS, TAF1, UBE2A, UPF3B, USP9X, ZC4H2, WNK3, ZMYM3, ZDHHC9, ZFX		
Intellektuelle Entwicklungsstörung und Makrozephalie *	4 - 6 Wo	E
Gen-Panel: ID131.03		
Intellektuelle Entwicklungsstörung und Makrozephalie: 48 Gene (147,3 kb)		
ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Makrozephalie: 21 Gene (70,8 kb)		
CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Makrozephalie: 10 Gene (20,2 kb)		
ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMCO1, ZBTB7A		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Makrozephalie: 18 Gene (57,9 kb)		
BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9		
Intellektuelle Entwicklungsstörung und Mikrozephalie *	4 - 6 Wo	E
Gen-Panel: ID129.02		
Intellektuelle Entwicklungsstörung und Mikrozephalie: 80 Gene (229,8 kb)		
ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNNB1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNPH2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Mikrozephalie: 32 Gene (100,5 kb)		
AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Mikrozephalie: 21 Gene (41,0 kb)		
ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Mikrozephalie: 27 Gene (88,3 kb)		
ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2		
Komplexe neurologische Entwicklungsstörungen (NED) *	4 - 6 Wo	E
Gen-Panel: ID358.01, 193 Gene (496,0 kb)		
ACBD6, ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BAZ2B, BCAS3, BPTF, BRAT1, C18ORF32, CAPRIN1, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CERT1, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DPH5, DYNC1I2, EEF1D, EMC10, ESAM, EXOC2, EXOC7, EXOC8, FAM177A1, FBXW11, FDFT1, FEM1B, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, GTPBP2, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HDAC4, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCND1, KCNN2, KDM5A, KDM6B, LNPK, MADD, MAP4K4, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NAV3, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPMT1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, RNU4-2, SARS1, SEC31A, SEL1L, SETD1A, SHMT2, SHQ1, SLC4A10, SMG8, SMG9, SMPD4, SNF8, SNIP1, SPOP, SPOUT1, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, TTI1, UBE3C, UBE4A, UBR5, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZBTB11, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6		

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Kabuki-Syndrom (KABUK) *, # Gen-Panel: ID127.00, 2 Gene (20,8 kb) KDM6A, KMT2D	3 - 5 Wo	E
Kleinwuchs, umfassende Diagnostik * Gen-Panel ID340.02 Kleinwuchs, umfassende Diagnostik: 207 Gene (511,0 kb) ACAN, ACP5, ACTB, ACTG1, AGPS, ALPL, AMMECR1, ANKRD11, ARCN1, ARSB, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BLM, BMP2, BMPR1B, BRAF, BRCA1, BRCA2, BRIP1, BTK, CBL, CCDC8, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CFAP410, COL10A1, COL11A2, COL27A1, COL2A, COMP, CREBBP, CRIPT, CSGALNACT1, CUL7, DDR2, DDRGK1, DHCR7, DNA2, DONSON, DPH1, EP300, ERCC4, ERCC6, ERCC8, EXOC6B, EXOSC2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGD1, FGFR1, FGFR3, FLNB, FN1, GALNS, GDF5, GH1, GHR, GHRHR, GHSR, GLB1, GLI2, GMNN, GNPAT, GPX4, GRHL2, GSC, GUSB, GZF1, HDAC8, HESX1, HMGA2, HRAS, HYAL1, IDUA, IGF1, IGF1R, IGF2, IGFALS, INSR, IRS1, IRS4, KIF22, KMT2A, KRAS, LARP7, LFNG, LHX3, LHX4, LTBP3, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MCM5, MRAS, NBAS, NBN, NEPRO, , NIPBL, NKX2-5, NKX3-2, NOTCH2, NPR2, NRAS, NSMCE2, OBSL1, ORC1, ORC4, ORC6, OTX2, PALB2, PAM16, PAPPA2, PAX8, PCNT, PEX5, PEX7, PHEX, PIK3R1, PISD, PLAG1, PLCB3, POC1A, POLR3GL, POP1, POU1F1, PPP1CB, PPP1R15B, PPP3CA, PRKG2, PRMT7, PROP1, PTH1R, PTPN11, PUS7, RAD21, RAD51, RAD51C, RAF1, RBBP8, RFWD3, RIT1, RMRP, RNPC3, RNU4ATAC, ROBO1, RPL13, RRAS2, RSPRY1, RTTN, SCUBE3, SGMS2, SHOC2, SHOX, SLC10A7, SLC26A2, SLX4, SMARCA2, SMC1A, SMC3, SOS1, SOS2, SOX, SOX3, SPRED2, SRCAP, STAT5B, TALDO1, TBCE, TBL1X, TBX15, TBX19, TBX2, THRA, TKT, TONSL, TOP3A, TRAIP, TRAPPC2, TRHR, TRIM37, TRIP11, TRMT10A , TRPV4, TSHB, TSHR, UBE2T, XRCC2, XRCC4 Wachstumshormonmangel (IGHD, CPHD): 14 Gene (23,0 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1, SOX3 Noonan-Syndrom (NS): 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Meier-Gorlin-Syndrom (MGORS): 8 Gene (12,6 kb) CDC45, CDC6, CDT1, GMNN, MCM5, ORC1, ORC4, ORC Seckel-Syndrom (SCKL): 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP Kongenitale Hypothyreose (CHNG): 8 Gene (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Skelettdysplasie (SED, SMD, AMD): 35 Gene (83,9 kb) ACAN, B3GALT6, BGN, BMPR1B, CFAP410, COL11A2, COL2A1, COMP, DDRGK1, DDR2, EXOC6B, FGFR3, FN1, GDF5, GPX4, KIF22, NEPRO, NKX3-2, NPR2, PAM16, PAPSS2, PISD, PLCB3, RMRP, RNU4ATAC, POP1, PRKG2, RPL13, RSPRY1, SIK3, SLC26A2, TONSL, TRAPPC2, TRIP11, TRPV4	4 - 6 Wo	E
Noonan-Syndrom (NS) * Gen-Panel: ID023.06, 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 Wo	E
Pitt-Hopkins-Syndrom (PTHS) * Gen-Panel: ID106.00, 3 Gene (10,6 kb) CNTNAP2, NRXN1, TCF4	3 - 5 Wo	E
Progerie und progeroide Syndrome * Gen-Panel: ID147.01, 25 Gene (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 Wo	E
RASopathien * Gen-Panel: ID015.05 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan-Syndrom (NS): 15 Gene (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 Wo	E
Rett-Syndrom (RTT) und ähnliche Krankheitsbilder * Gen-Panel: ID125.01, 16 Gene (45,3 kb) CDKL5, FOXG1, GABBR2, GABRA2, GABRB2, GRIN2B, HTT, IQSEC2, MECP2, MEF2C, NTNG2, SHANK3, STXBP1, TCF4, UBE3A, WDR45	3 - 5 Wo	E
Rubinstein-Taybi-Syndrom (RSTS) * Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
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
Sprachentwicklungsstörung (DLD, CAS) * Gen-Panel: ID368.00 Sprachentwicklungsstörung (DLD, CAS): 34 Gene (119,7 kb) ATP2C2, BCL11A, BUD13, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, NFXL1, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TM4SF20, TNRC6B, UPF2, WDR5, ZFHX4, ZNF142, ZNF277 Kindliche Sprechapraxie (CAS): 29 Gene (110,2 kb) BCL11A, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TNRC6B, UPF2, WDR5, ZFHX4, ZNF142 Spezifische Sprachbeeinträchtigung (SLI): 5 Gene (9,5 kb) TP2C2, BUD13, NFXL1, TM4SF20, ZNF277	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
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: ID391.01, 60 Gene (142,0 kb) ADAD2, ADGRG2, AR, C14ORF39, CATIP, CATSPER1, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, FANCM, FKBP6, GCNA, HFM1, KASH5, KCTD19, KLHL10, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PMFBP1, PNLDC1, RAD21L1, RBBP7, RNF212, RPL10L, SHOC1, SOHLH1, SPATA22, SPINK2, SPO11, STAG3, STRA8, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TERB1, TERB2, TEX11, TEX14, TEX15, USP9Y, XRCC2, ZMYND15, ZSWIM7	4 - 6 Wo	E
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) * Gen-Panel: ID170.05, 40 Gene (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 Wo	E
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 ID192.07 Männliche Infertilität, umfassende Diagnostik: 135 Gene (446,9 kb) ACR, ACTL7A, ACTL9, ADAD2, ADGRG2, AK7, AK9, AKAP3, AR, ARMC12, ARMC2, ATG4D, AURKC, BRDT, C14ORF39, CATIP, CATSPER1, CATSPER2, CATSPERT, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP206, CFAP251, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH12, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, GPAT2, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRRC23, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PLD6, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RAD21L1, RBBP7, RNF212, RPL10L, SEPTIN4, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG6, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SPO11, SSX1, STAG3, STK33, STRA8, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TDRD12, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Störung der Spermatogenese (SPGF): 112 Gene (395,3 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, ATG4D, AURKC, BRDT, C14ORF39, CATIP, CATSPER1, CATSPERT, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP206, CFAP251, CT55, CYLC1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH12, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, IFT74, IQCN, KASH5, KCNU1, KLHL10, LRRC23, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RBBP7, RNF212, RPL10L, SEPTIN4, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Obstruktive Azoospermie (CBAVD, OAZON): 3 Gene (8,2 kb) ADGRG2, CFTR, CLDN2	4 - 6 Wo	E
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 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
Variante der Geschlechtsentwicklung (DSD) * Gen-Panel ID117.04 Variante der Geschlechtsentwicklung (DSD): 55 Gene (106,2 kb) AKR1C2, AKR1C4, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR3C1, NR5A1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX3, SOX8, SOX9, SOX10, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 20 Gene (36,1 kb) ANOS1, AKR1C2, AKR1C4, AR, CBX2, DHH, DHX37, DMRT1, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 7 Gene (8,7 kb) NR2F2, NR5A1, SOX3, SOX9, SRY, WT1, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 41 Gene (80,2 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, NR3C1, POR, PPP1R12A, PPP2R3C, RPL10, RSPO1, SAMD9, SGPL1, SOX9, SOX10, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
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) *, # Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
Cholestase, umfassende Diagnostik * 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	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
Intrahepatische Cholestase (PFIC, CBAS) * 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	3 - 5 Wo	E
Kolorektales Karzinom * Gen-Panel: ID006.10, 24 Gene (66,3 kb) APC, ATM, AXIN2, BAP1, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53	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
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
Morbus Hirschsprung (HSCR) * 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	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
Pankreatitis (PCTT) * 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	3 - 5 Wo	E
Polyposis-Syndrom (PS, FAP) * Gen-Panel: ID005.07, 16 Gene (41,6 kb) APC, AXIN2, BAP1, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	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
Viszerale Myopathien und Neuropathien, umfassende Diagnostik * 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	3 - 5 Wo	E

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: ID392.01 Anämien, umfassende Diagnostik: 188 Gene (357,1 kb) ABCB7, ABCG5, ABCG8, ADA, ADA2, ADAMTS13, ADH5, AK1, AK2, ALAD, ALAS2, ALDH2, ALDOA, AMN, ANK1, ATM, ATP11C, ATRX, BLM, BRCA1, BRCA2, BRIP1, C3, C1GALT1C1, CBLIF, CD46, CD59, CDAN1, CDIN1, CFB, CFH, CFHR1, CFHR3, CFI, COL4A1, COX4I2, CPOX, CUBN, CYB5R3, DGKE, DHFR, DKC1, DNASE2, DNAJC21, ELANE, EPAS1, EPB41, EPB42, EPO, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FOXP3, FTCD, FTL, G6PC3, G6PD, GATA1, GCLC, GLRX5, GPI, GPX1, GSR, GSS, HAMP, HBA1, HBA2, HBB, HBD, HBG1, HBG2, HEATR3, HFE, HK1, HSPA9, HSCB, IARS2, IREB2, KCNN4, KIF23, KLF1, LARS2, LCAT, LIG4, LPIN2, MAD2L2, MDM4, MMACHC, MMADHC, MPIG6B, MPL, MTHFD1, MTR, MTRR, MYSM1, NBN, NDUFB11, NHEJ1, NHLRC2, NHP2, NT5C3A, PALB2, PFKM, PGK1, PIEZO1, PKLR, PRF1, PUS1, RACGAP1, RAD51, RAD51C, RBSN, REN, RFWD3, RHAG, RMRP, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RUNX1, SBDS, SEC23B, SH2B3, SH2D1A, SLC11A2, SLC19A1, SLC19A2, SLC25A38, SLC30A7, SLC40A1, SLC46A1, SLC2A1, SLC4A1, SLX4, SMAD4, SPTA1, SPTB, SRP72, STAT3, STEAP3, STIM1, TBXAS1, TCN2, TERC, TERT, TF, TFR2, THBD, THPO, TINF2, TMPRSS6, TOMM70, TP53, TPI1, TRNT1, TSR2, UMPS, UBE2T, VPS4A, WRAP53, XK, XRCC2, YARS2 Diamond-Blackfan-Anämie (DBA): 22 Gene (11,5 kb) HEATR3, RPL5, RPL9, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Sideroblastische Anämie (SIDBA): 10 Gene (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2 Megaloblastische Anämie: 13 Gene (31,4 kb) AMN, CUBN, DHFR, FTCD, MMADHC, MTHFD1, MTR, MTRR, SLC19A1, SLC19A2, SLC46A1, TCN2, UMPS Kongenitale nicht-sphärozytäre hämolytische Anämie (CNSHA): 10 Gene (15,3 kb) AK1, G6PD, GATA1, GCLC, GPI, GSR, GSS, HK1, NT5C3A, PKLR Kongenitale dyserythropoetische Anämie (CDAN): 6 Gene (12,7 kb) CDAN1, CDIN1, KIF23, KLF1, RACGAP1, SEC23B Sphärozytose (SPH) und Elliptozytose (EL): 6 Gene (26,6 kb) ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB Hereditäre Stomatozytose: 8 Gene (20,8 kb) ABCB6, ABCG5, ABCG8, KCNN4, PIEZO1, RHAG, SLC2A1, SLC4A1	4 - 6 Wo	E
Atypisches hämolytisch-urämisches Syndrom (AHUS) * Gen-Panel: ID163.04, 20 Gene (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 Wo	E
Diamond-Blackfan-Anämie (DBA) * Gen-Panel: ID394.00, 21 Gene (11,9 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL17, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2	3 - 5 Wo	E
Erythrozytose (ECYT) * Gen-Panel: ID138.02, 11 Gene (13,9 kb) BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL	3 - 5 Wo	E
Fanconi-Anämie (FANC) * Gen-Panel: ID043.02, 21 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E
Hämophilie * Gen-Panel: ID154.01, 5 Gene (18,9 kb) F8, F9, LMAN1, MCFD2, VWF	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS) * Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
Knochenmarkversagen (BMF) * Gen-Panel: ID357.01 Knochenmarkversagen (BMF): 28 Gene (47,6 kb): ACD, ADH5, ALDH2, DCLRE1B, DKC1, DNAJC21, DUT, EFL1, ERCC6L2, MDM4, MYSM1, NAF1, NHP2, NOP10, PARN, POT1, RPA1, RTEL1, SBDS, SLC30A7, SRP72, TERC, TERT, TINF2, TP53, TYMS, WRAP53, ZCCHC8 Knochenmarkinsuffizienz-Syndrom (BMFS): 10 Gene (18,0 kb) ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, SLC30A7, SRP72, TP53 Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 9 Gene (16,6 kb) NAF1, NOP10, PARN, POT1, RPA1, RTEL1, TERC, TERT, ZCCHC8 Dyskeratosis congenita: 12 Gene (18,2 kb) ACD, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53	3 - 5 Wo	E
Neutropenie * Gen-Panel: ID189.03 Neutropenie: 33 Gene (55,6 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SEC61A1, SLC37A4, SMARCD2, SRP54, SRP68, TAFAZZIN, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Schwere kongenitale Neutropenie (SCN): 13 Gene (19,7 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SEC61A1, SRP54, SRP68, TCIRG1, VPS45, WAS Syndrome mit Neutropenie: 22 Gene (38,8 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, TAFAZZIN, USB1, VPS13B, WDR1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hämatologische Erkrankungen		
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
Porphyrie * Gen-Panel: ID153.01, 10 Gene (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 Wo	E
Sideroblastische Anämie (SIDBA) * Gen-Panel: ID355.00, 10 Gene (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2	3 - 5 Wo	E
Sphärozytose (SPH) und Elliptozytose (EL) * 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	3 - 5 Wo	E
Thrombophilie (THPH) * Gen-Panel: ID150.01, 12 Gene (25,2 kb) F2, F5, F9, F13B, HABP2, HRG, MTHFR, PROC, PROS1, SERPINC1, SERPIND1, THBD	3 - 5 Wo	E
Thrombozytendefekte, umfassende Diagnostik * 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	4 - 6 Wo	E
Thrombozytopenie (THC) * 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	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hämatologische Erkrankungen		
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		
Haut- und Zahnerkrankungen		
Adams-Oliver-Syndrom (AOS) *	3 - 5 Wo	E
Gen-Panel: ID259.00, 6 Gene (23,2 kb)		
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		
Albinismus, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel: ID175.05		
Albinismus, umfassende Diagnostik: 33 Gene (73,8 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1		
Okulokutaner Albinismus (OCA, OA): 9 Gene (13,2 kb)		
DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Waardenburg-Syndrom (WS): 8 Gene (9,3 kb)		
EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR		
Griscelli-Syndrom (GS): 3 Gene (8,0 kb)		
MLPH, MYO5A, RAB27A		
Amelogenesis imperfecta (AI) *	3 - 5 Wo	E
Gen-Panel: ID232.01, 17 Gene (29,4 kb)		
ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAPH, RELT, SLC24A4, WDR72		
Cowden-Syndrom (CWS) *	3 - 5 Wo	E
Gen-Panel: ID075.01, 8 Gene (12,8 kb)		
AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1		
Cutis laxa (ARCL, ADCL) *	3 - 5 Wo	E
Gen-Panel: ID109.03, 13 Gene (32,1 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1, RIN2		
Dyskeratosis congenita (DKC) *	3 - 5 Wo	E
Gen-Panel: ID347.01, 15 Gene (24,1 kb)		
ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53		
Ektodermale Dysplasie (ECTD) *	3 - 5 Wo	E
Gen-Panel: ID136.04		
Ektodermale Dysplasie (ECTD): 44 Gene (74,6 kb)		
AP1B1, CDH3, CHUK, CST6, DLX3, DSG4, EDA, EDAR, EDARADD, GJA1, GJB2, GJB6, GRHL2, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LRP6, MSX1, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, WNT10A		
Ektodermale Dysplasie (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, C3ORF52, CDH1, CDH3, CDSN, CHUK, CST6, CTNND1, CTSK, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, EVC, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB2, GJB6, GRHL2, HEPHL1, HOXC13, HR, IFT122, IFT140, IFT43, IFT52, IKBKG, INSR, KCTD1, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LIPH, LPAR6, LRP6, MBTPS2, MSX1, NECTIN1, NECTIN4, NFKB2, NFKBIA, NLRP1, PAX9, PEX1, PEX6, PKP1, PORCN, PRKD1, RIPK4, ROGDI, RSPO4, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SNRPE, SREBF1, ST14, TBC1D24, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, UBR1, WDR19, WDR35, WNT10A		
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), nicht-syndromal: 20 Gene (27,5 kb)		
CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A		
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), syndromal: 28 Gene (51,0 kb)		
AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2		
Komplexe Syndrome mit ektodermaler Dysplasie: 42 Gene (120,5 kb)		
ANTXR1, ARID1A, ARID1B, ATP6V1B2, CDH1, CTNND1, CTSK, DSG4, DSP, EVC, EVC2, FGF10, FGFR2, FGFR3, HEPHL1, IFT122, IFT140, IFT43, IFT52, INSR, KCTD1, KRT14, KRT16, KRT17, KRT81, KRT83, KRT86, NLRP1, PEX1, PEX6, ROGDI, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SREBF1, TBC1D24, UBR1, WDR19, WDR35		

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Epidermolysis bullosa (EB) * 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	4 - 6 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
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: ID265.01, 14 Gene (40,2 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TBC1D32, TCTN3, TMEM107, ZRSR2	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
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-Syndrom * Gen-Panel: ID294.01, 34 Gene (73,7 kb) AMER1, ARCN1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, GNAI3, MYMK, MYMX, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Palmoplantarkeratose (PPK) *	3 - 5 Wo	E
Gen-Panel: ID323.00		
Palmoplantarkeratose (PPK): 33 Gene (58,2 kb)		
AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB3, GJB4, JUP, KDSR, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT10, KRT14, KRT16, KRT17, KRT83, LORICRIN, MBTPS2, PERP, RHBDF2, SERPINB7, SLURP1, SMARCAD1, TAT, TRPM4, TRPV3, WNT10A		
Palmoplantarkeratose (PPK), nicht-syndromal: 11 Gene (25,7 kb)		
AAGAB, AQP5, DSG1, DSP, KRT1, KRT6C, KRT9, KRT10, KRT16, SERPINB7, TRPV3		
Palmoplantarkeratose (PPK), syndromal: 15 Gene (22,4 kb)		
CTSC, ENPP1, GJA1, GJB2, KRT14, MBTPS2, JUP, LORICRIN, PERP, RHBDF2, SLURP1, SMARCAD1, TAT, TRPV3, WNT10A		
Erythrokeratodermia variabilis et progressiva (EKVP): 7 Gene (9,5 kb)		
GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4		
Pachyonychia congenita (PC): 4 Gene (6,1 kb)		
KRT6A, KRT6B, KRT16, KRT17		
Primäres Lymphödem (LMPHM) *	3 - 5 Wo	E
Gen-Panel: ID372.00, 21 Gene (69,4 kb)		
ADAMTS3, ANGPT2, CALCRL, CCBE1, CELSR1, EPHB4, ERG, FAT4, FLT4, FOXC2, GATA2, GJA1, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, THSD1, TIE1, VEGFC		
Progerie und progeroide Syndrome *	3 - 5 Wo	E
Gen-Panel: ID147.01, 25 Gene (61,9 kb)		
ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24		
Selektive Zahn-Agenesie (STHAG) *	3 - 5 Wo	E
Gen-Panel: ID151.02, 16 Gene (26,9 kb)		
EDA, EDAR, EDARADD, GREM2, IRF6, KDF1, KREMEN1, LRP6, LTBP3, MSX1, PAX9, PTH1R, TP63, TSPEAR, WNT10A, WNT10B		
Systemerkrankungen mit multiplen Café-au-lait-Flecken *	3 - 5 Wo	E
Gen-Panel ID351.00		
Systemerkrankungen mit multiplen Café-au-lait-Flecken: 9 Gene (27,6 kb)		
BRAF, MLH1, MSH2, MSH6, NF1, PMS2, PTPN11, RAF1, SPRED1		
Konstitutionelle Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb)		
MLH1, MSH2, MSH6, PMS2		
Neurofibromatose (NF1, NFLS): 2 Gene (9,8 kb)		
NF1, SPRED1		
LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb)		
BRAF, PTPN11, RAF1		
Tuberöse Sklerose (TSC) *	2 - 4 Wo	E
Gen-Panel: ID332.00, 2 Gene (8,9 kb)		
TSC1, TSC2		
Xeroderma pigmentosum (XP) *	3 - 5 Wo	E
Gen-Panel: ID282.00, 10 Gene (23,5 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC		
Herz- und Gefäßerkrankungen		
Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC) *	4 - 6 Wo	E
Gen-Panel: ID010.03, 27 Gene (185,3 kb)		
ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN		
Atriumseptumdefekt und Ventrikelseptumdefekt (ASD, VSD, AVSD) *	3 - 5 Wo	E
Gen-Panel: ID143.00, 12 Gene (21,1 kb)		
ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID137.06		
Bindegewebserkrankungen (EDS, MFS, LDS), umfassende Diagnostik: 85 Gene (283,3 kb)		
ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, ALDH18A1, ASPH, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, DCC, DLG4, DSE, EFEMP1, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GORAB, IPO8, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NKAP, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THBS2, THSD4, TNXB, ZNF469		
Marfan-Syndrom (MFS): 3 Gene (11,8 kb)		
FBN1, TGFBR2, TGFBR1		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 18 Gene (45,7 kb)		
ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Cutis laxa-Syndrom (ARCL, ADCL): 12 Gene (29,4 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
Stickler-Syndrom (STL): 6 Gene (22,0 kb)		
COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2		
Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Bikuspide Aortenklappe (AOVD) *	3 - 5 Wo	E
Gen-Panel: ID301.00, 6 Gene (16,7 kb)		
GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2		
Brugada-Syndrom (BRGDA) *	3 - 5 Wo	E
Gen-Panel: ID014.02, 23 Gene (56,4 kb)		
ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4		
CADASIL und CARASIL *, #	3 - 5 Wo	E
Gen-Panel: ID167.01, 3 Gene (9,4 kb)		
HTRA1, NOTCH3, TREX1		
CHARGE-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID307.00, 3 Gene (12,9 kb)		
CHD7, SEMA3E, TBX22		
Cutis laxa (ARCL, ADCL) *	3 - 5 Wo	E
Gen-Panel: ID109.03, 13 Gene (32,1 kb)		
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP1, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1, RIN2		
Dilatative Kardiomyopathie (CMD, DCM) *	4 - 6 Wo	E
Gen-Panel: ID008.05, 68 Gene (301,9 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1		
Ehlers-Danlos-Syndrom (EDS) *	3 - 5 Wo	E
Gen-Panel: ID039.06		
Ehlers-Danlos-Syndrom (EDS): 21 Gene (83,1kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, THBS2, TNXB, ZNF469		
Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 9 Gene (39,6 kb)		
C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1, THBS2		
Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb)		
ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469		
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		
Fallot-Tetralogie (TOF) *	3 - 5 Wo	E
Gen-Panel: ID144.01, 12 Gene (23,5 kb)		
FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2		
Familiäres Vorhofflimmern (ATFB) *	3 - 5 Wo	E
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		
Frühes Repolarisationssyndrom (ERS) *	3 - 5 Wo	E
Gen-Panel: ID330.00, 12 Gene (38,9 kb)		
ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A		
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		

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
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
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.08 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 40 Gene (117,6 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (12,6 kb) BGN, IPO8, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit thorakalem Aortenaneurysma: 31 Gene (96,5 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, IPO8, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E
Long-QT-Syndrom (LQT) * Gen-Panel: ID013.01, 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN	3 - 5 Wo	E
Kardiale Arrhythmien, umfassende Diagnostik * Gen-Panel: ID026.03 Kardiale Arrhythmien, umfassende Diagnostik: 71 Gene (274,2 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, EMD, FGF12, GJA5, GNB2, GNB5, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYH6, MYL4, NKX2-5, NPPA, NUP155, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TANGO2, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN Long-QT-Syndrom (LQT): 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN Brugada-Syndrom (BRGDA): 23 Gene (56,4 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPDL, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2V, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4 Frühes Repolarisationssyndrom (ERS): 12 Gene (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A Familiäres Vorhofflimmern (ATFB): 17 Gene (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A Short-QT-Syndrom (SQT): 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCL4A3 Sick-Sinus-Syndrom (SSS): 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN Arrhythmogene rechtsventrikuläre Dysplasie (ARVD): 15 Gene (150,6 kb) CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, PLN, LMNA, PKP2, PRKAG2, RYR2, TGFB3, TMEM43, TTN	4 - 6 Wo	E
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) * Gen-Panel: ID012.03, 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Kardiomyopathien, umfassende Diagnostik * Gen-Panel: ID027.05 Kardiomyopathien, umfassende Diagnostik: 154 Gene (487,0 kb) ABCC9, ACTA1, ACTC1, ACTN2, ALPK3, ANKRD1, APOA1, B2M, BAG3, BAG5, BRAF, CACNA1C, CACNB2, CALR3, CAP2, CASQ2, CAV3, CDH2, COA5, COA6, CORIN, COX15, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DMPK, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGA, FHL1, FHL2, FHOD3, FKRP, FKTN, FLII, FLNC, FTH1, FXN, GAA, GATA4, GATAD1, GET3, GLA, GSN, HADHA, HAMP, HCN4, HFE, HJV, HRAS, ILK, JPH2, JUP, KCNQ1, KIF20A, KLF10, KLHL24, KRAS, KY, LAMA4, LAMP2, LDB3, LIMS2, LMNA, LMOD2, LRRC10, LYZ, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM10, MIB1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOT, MYOZ2, MYPN*, NEBL, NEXN, NKX2-5, NNT, NONO, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, PYROXD1, RAF1, RBM20, RIT1, RPL3L, RRAS2, RYR2, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC40A1, SLC25A4, SOS1, SOS2, SPRED2, SVIL, SYNE1, SYNE2, TAFAZZIN, TBX5, TBX20, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TRPM4, TTN, TTR, UNC45B, VCL, VEZF1 Dilatative Kardiomyopathie (DCM, CMD): 68 Gene (301,9 kb) ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1 Hypertrophe Kardiomyopathie (HCM, CMH): 56 Gene (253,8 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL Restriktive Kardiomyopathie (RCM): 15 Gene (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC): 27 Gene (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN Nichtdilatierte linksventrikuläre Kardiomyopathie (NDLVC, LVNC): 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
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
Konotrunkale Herzfehlbildung (CTHM) * Gen-Panel: ID160.02, 18 Gene (42,8 kb) FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, JAG1, NKX2-5, NKX2-6, NOTCH1, NR2F2, SMAD2, PLXND1, TAB2, TBX1, TMEM260, ZFPM2, ZIC3	3 - 5 Wo	E
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 ID194.06 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 50 Gene (157,9 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP1, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, IPO8, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 Loeys-Dietz-Aortenaneurysma-Syndrom (LDS): 17 Gene (42,1 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Stickler-Syndrom (STL): 6 Gene (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Syndrome mit marfanoidem Habitus: 20 Gene (52,2 kb) CBS, DLG4, EFEMP1, EFEMP2, FBN1, FBN2, NKAP, NPR2, MED12, PLOD1, PRDM5, SKI, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, UPF3B, ZDHHC9	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Multipler kongenitaler Herzdefekt (CHTD) *	3 - 5 Wo	E
Gen-Panel: ID302.01, 9 Gene (21,5 kb)		
FLT4, GATA5, GDF1, NR2F2, PLD1, PLXND1, SMAD2, TAB2, ZIC3		
Muskelerkrankungen mit Herzbeteiligung *	4 - 6 Wo	E
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		
Nichtdilatierte linksventrikuläre Kardiomyopathie (NDLVC, LVNC) *	4 - 6 Wo	E
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		
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		
Plötzlicher Herztod *	4 - 6 Wo	E
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		
Pulmonale Hypertonie (PAH, PPH) *	3 - 5 Wo	E
Gen-Panel: ID281.01, 23 Gene (49,9 kb)		
ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4		
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		
Restriktive Kardiomyopathie (RCM) *	4 - 6 Wo	E
Gen-Panel: ID105.03, 15 Gene (135,7 kb)		
ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR		
Short-QT-Syndrom (SQT) *	3 - 5 Wo	E
Gen-Panel: ID233.01, 7 Gene (22,1 kb)		
CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3		
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		

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Sick-Sinus-Syndrom (SSS) * Gen-Panel: ID107.01, 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) * Gen-Panel: ID020.02 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 17 Gene (44,1 kb) ACTA2, COL3A1, FBN1, FOXE3, IPO8, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	3 - 5 Wo	E
Viszerale Heterotaxie (HTX) * Gen-Panel: ID145.01, 18 Gene (72,4 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, DNAAF1, DNAH5, DNAH9, DNAH11, MMP21, MNS1, NODAL, ODAD2, PKD1L1, ZIC3	3 - 5 Wo	E
Zerebrale Kleingefäßerkrankung (BSVD) * Gen-Panel: ID325.00, 3 Gene (12,0 kb) COL4A1, COL4A2, COLGALT1	3 - 5 Wo	E
Zerebrovaskuläre Erkrankungen und Schlaganfall * Gen-Panel: ID234.02 Zerebrovaskuläre Erkrankungen und Schlaganfall: 44 Gene (130,0 kb) ACE, ADA2, ACTA2, ALOX5AP, APOE, APP, CBS, CCM2, COL3A1, COL4A1, COL4A2, COLGALT1, CST3, F2, F5, FBN1, FLNA, GAA, GLA, GSN, GUCY1A1, HTRA1, ITM2B, JAG1, KRIT1, MTHFR, MYH11, MYLK, NOS3, NOTCH3, OTC, PDCD10, POLG, PRKCH, PRNP, RNF213, SLC2A10, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, TREX1, TTR Zerebrale Amyloidangiopathie: 6 Gene (7,1 kb) APP, CST3, GSN, ITM2B, PRNP, TTR Zerebrale Kleingefäßerkrankung (BSVD): 3 Gene (12,0 kb) COL4A1, COL4A2, COLGALT1 Zerebrale kavernöse Fehlbildung (CCM): 3 Gene (4,2 kb) CCM2, KRIT1, PDCD10 Zerebrales Aneurysma und Dissektion: 10 Gene (32,8 kb) ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Moyamoya-Erkrankung (MYMY): 4 Gene (22,2 kb) ACTA2, GUCY1A1, JAG1, RNF213 CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3	4 - 6 Wo	E
HNO-Erkrankungen		
Branchiootorenales Syndrom (BOR) * Gen-Panel: ID315.00, 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 Wo	E
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) * Gen-Panel: ID170.05, 40 Gene (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 Wo	E
Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik * Gen-Panel: ID237.03 Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik: 133 Gene (374,8 kb) ABCC1, ACTG1, ADCY1, AFG2B, AIFM1, ATOH1, ATP11A, ATP2B2, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GPRASP2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MAP1B, MARVELD2, MCM2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PI4KB, PJVK, PKHD1L1, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, THOC1, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, USP48, WBP2, WFS1, WHRN Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA): 60 Gene (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24*, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1 Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB): 83 Gene (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX): 6 Gene (11,8 kb) AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX	4 - 6 Wo	E

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

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		
Knochenmarkversagen (BMF) *	3 - 5 Wo	E
Gen-Panel: ID357.01		
Knochenmarkversagen (BMF): 28 Gene (47,6 kb):		
ACD, ADH5, ALDH2, DCLRE1B, DKC1, DNAJC21, DUT, EFL1, ERCC6L2, MDM4, MYSM1, NAF1, NHP2, NOP10, PARN, POT1, RPA1, RTEL1, SBDS, SLC30A7, SRP72, TERC, TERT, TINF2, TP53, TYMS, WRAP53, ZCCHC8		
Knochenmarkinsuffizienz-Syndrom (BMFS): 10 Gene (18,0 kb)		
ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, SLC30A7, SRP72, TP53		
Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 9 Gene (16,6 kb)		
NAF1, NOP10, PARN, POT1, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Dyskeratosis congenita: 12 Gene (18,2 kb)		
ACD, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53		
Neutropenie *	3 - 5 Wo	E
Gen-Panel: ID189.03		
Neutropenie: 33 Gene (55,6 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SEC61A1, SLC37A4, SMARCD2, SRP54, SRP68, TAFAZZIN, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1		
Schwere kongenitale Neutropenie (SCN): 13 Gene (19,7 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SEC61A1, SRP54, SRP68, TCIRG1, VPS45, WAS		
Syndrome mit Neutropenie: 22 Gene (38,8 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, TAFAZZIN, USB1, VPS13B, WDR1		
Schwerer kombinierter Immundefekt (SCID) *	3 - 5 Wo	E
Gen-Panel: ID381.00, 21 Gene (47,6 kb)		
ADA, AK2, BCL11B, CD3E, CD3D, CIITA, CORO1A, DCLRE1C, FOXN1, IL2RG, IL7R, LAT, LIG4, JAK3, NHEJ1, POLD3, PRKDC, PTPRC, RAC2, RAG1, RAG2		
Periodische Fiebersyndrome *	3 - 5 Wo	E
Gen-Panel: ID088.04, 12 Gene (26,3 kb)		
ELANE, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSTPIP1, RIPK1, TNFRSF1A		

Erkrankung/Diagnostik	Dauer	Material
Immunologische Erkrankungen		
Primäre Immundefekte, umfassende Diagnostik 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	4 - 6 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) * Gen-Panel: ID251.01, 10 Gene (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 Wo	E
Hydrozephalus * 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	3 - 5 Wo	E
Intellektuelle Entwicklungsstörung und Makrozephalie * 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	4 - 6 Wo	E
Intellektuelle Entwicklungsstörung und Mikrozephalie * 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	4 - 6 Wo	E
Mikrozephalie, umfassende Diagnostik * 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	4 - 6 Wo	E

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		
Coenzym-Q10-Mangel (COQ10D) * Gen-Panel: ID225.01, 15 Gene (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2	3 - 5 Wo	E
Defizienz der mitochondrialen Komplexe I bis V (MCDN) * Panel: ID074.03 Defizienz der mitochondrialen Komplexe I bis V (MCDN): 83 Gene (60,7 kb) ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX5A, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, CYC1, DNAJC30, FOXRED1, LRPPRC, LYRM7*, MTFMT, NDUFA1, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, PET100, PET117, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SURF1, TACO1, TIMMDC1, TMEM70, TMEM126B, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ Mitochondrialer Komplex-I-Mangel, kernkodierter Typ (MC1DN): 39 Gene (29,3 kb) ACAD9, DNAJC30, FOXRED1, MTFMT, NDUFA1, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, TIMMDC1, TMEM126B Mitochondrialer Komplex-II-Mangel, kernkodierter Typ (MC2DN): 4 Gene (3,7 kb) SDHA, SDHAF, SDHB, SDHD Mitochondrialer Komplex-III-Mangel, kernkodierter Typ (MC3DN): 10 Gene (7,1 kb) BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1 Mitochondrialer Komplex-IV-Mangel, kernkodierter Typ (MC4DN): 23 Gene (15,8 kb) COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, COX5A, LRPPRC, NDUFA4, PET100, PET117, SCO1, SCO2, SURF1, TACO1 Mitochondrialer Komplex-V-(ATP-Synthase-)Mangel, kernkodierter Typ (MC5DN): 7 Gene (4,8 kb) ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, TMEM70	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Mitochondriopathien		
Kombinierter oxidativer Phosphorylierungsmangel (COXPD) *	3 - 5 Wo	E
Gen-Panel: ID287.01, 60 Gene (80,3 kb)		
AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2		
Lebersche hereditäre Optikusneuropathie (LHON) #	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 #	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: ID324.01, 21 Gene (29,3 kb)		
AGK, DGUOK, FBXL4, GUK1, LIG3, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP		
Mitochondriale Myopathie / Enzephalopathie #	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 #	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) * Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
Alport-Syndrom (ATS) * Gen-Panel: ID099.00, 4 Gene (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 Wo	E
Bardet-Biedl-Syndrom (BBS) * Gen-Panel: ID093.02, 21 Gene (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 Wo	E
CHARGE-Syndrom * Gen-Panel: ID307.00, 3 Gene (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 Wo	E
Coffin-Siris-Syndrom (CSS) * Gen-Panel: ID118.02, 14 Gene (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 Wo	E
Cornelia-de-Lange-Syndrom (CDLS) * Gen-Panel: ID033.02, 8 Gene (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 Wo	E
Fehlbildungssyndrome mit überwiegend fazialer Beteiligung * Gen-Panel: ID279.00 Fehlbildungssyndrome mit überwiegend fazialer Beteiligung: 25 Gene (90,9 kb) C2CD3, COL11A2, CPLANE1, DDX59, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GRIP1, IFT57, INTU, KIAA0753, MEGF8, MYH3, MYMK, MYT1, OFD1, RAB23, RBM10, TCTN3, TGDS, TMEM107, TNNI2, TWIST1 Akrozephalosyndaktylie-Syndrom (ACS): 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107 Fraser-Syndrom (FRASRS): 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1 Pierre-Robin-Syndrom: 4 Gene (9,7 kb) COL11A2, MYMK, RBM10, TGDS	3 - 5 Wo	E
FG-Syndrom (FGS) * Gen-Panel: ID215.00, 3 Gene (17,2 kb) CASK, FLNA, MED12	3 - 5 Wo	E
Kabuki-Syndrom (KABUK) *, # Gen-Panel: ID127.00, 2 Gene (20,8 kb) KDM6A, KMT2D	3 - 5 Wo	E
Kombinierter oxidativer Phosphorylierungsmangel (COXPD) * Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 Wo	E
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
Marfan-Syndrom (MFS) *, # Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2	2 - 4 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
Orofaziodigitales Syndrom (OFD) * Gen-Panel: ID265.01, 14 Gene (40,2 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TBC1D32, TCTN3, TMEM107, ZRSR2	3 - 5 Wo	E
Rubinstein-Taybi-Syndrom (RSTS) * Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E

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
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
Neurodegenerative Erkrankungen		
Alzheimer-Krankheit (AD) * Gen-Panel: ID157.02, 9 Gene (23,4 kb) A2M, ABCA7, ADAM10, APOE, APP, NOS3, PSEN1, PSEN2, TREM2	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
CADASIL und CARASIL *, # Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E
Choreatiforme Bewegungsstörungen * 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	3 - 5 Wo	E
Dystonie (DYT) * 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	3 - 5 Wo	E
Episodische Ataxie (EA) * Gen-Panel: ID184.02, 6 Gene (20,6 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3	3 - 5 Wo	E
Essentieller Tremor (ETM) * Gen-Panel: ID195.01, 4 Gene (16,6 kb) DRD3, FUS, SCN4A, TENM4	3 - 5 Wo	E
Familiäres episodisches Schmerzsyndrom (FEPS) * Gen-Panel: ID268.01, 6 Gene (22,3 kb) GLA, SCN9A, SCN10A, SCN11A, TRPA1, TTR	3 - 5 Wo	E
Frontotemporale Demenz (FTD) * 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	3 - 5 Wo	E
Hereditäre sensorische und autonome Neuropathie (HSAN, HSN) * Gen-Panel: ID086.02, 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Hereditäre Ataxien, umfassende Diagnostik * 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	4 - 6 Wo	E
Hereditäre Neuropathien (HMSN, HSAN, SMA), umfassende Diagnostik * Gen-Panel ID374.02 Hereditäre Neuropathien (HMSN, HSAN, SMA), umfassende Diagnostik: 245 Gene (598,6 kb) AAAS, AARS1, ABCA1, ABCD1, ABHD12, ACOX1, ADA2, ADCY6, ADGRG6, ADPRS, AGTPBP1, AGXT, AIFM1, AMACR, AP1S1, AP5Z1, APOA1, APTX, AR, ARHGEF10, ARL6IP1, ARSA, ASAH1, ASCC1, ATAD3A, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP7A, ATXN10, ATXN1, ATXN2, ATXN3, ATXN7, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CADM3, CAPN1, CD59, CFAP276, CHCHD10, CLP1, CNTNAP1, COA7, COQ7, COX20, COX6A1, CPOX, CTDP1, CYP27A1, CYP2U1, DARS2, DCTN1, DHH, DHTKD1, DHX9, DMXL2, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, ERCC6, ERCC8, ETFDH, EXOSC3, FA2H, FAH, FBLN5, FBXO38, FDXR, FGD4, FICD, FIG4, FLVCR1, FMR1, FXN, GALC, GAN, GARS1, GBA2, GBF1, GDAP1, GJB1, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HPDL, HSPB1, HSPB3, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, LYST, MAG, MARS1, MCM3AP, MED25, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MTTP, MYH14, NAGA, NAGLU, NARS1, NDC1, NDRG1, NDUFS6, NEFH, NEFL, NEMF, NFASC, NGF, NTRK1, NUDT2, OPA1, OPA3, PDHA1, PDK3, PDXK, PDYN, PEX10, PEX7, PHYH, PIEZO2, PIGB, PLA2G6, PLAAT3, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, PNPT1, POLG, POLR3A, POLR3B, PPOX, PRDM12, PRNP, PRPS1, PRX, PTRH2, RAB7A, REEP1, RETREG1, RTN2, SACS, SAMD9L, SARS1, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SMN1, SMN2, SORD, SOX10, SPAST, SPG11, SPG7, SPTAN1, SPTBN4, SPTLC1, SPTLC2, SURF1, SYT2, TBCE, TECPR2, TFG, TRIM2, TRIP4, TRMT5, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, UCHL1, VAPB, VCP, VPS13D, VRK1, VWA1, WARS1, WNK1, XK, XPA, YARS1, ZFYVE26	4 - 6 Wo	E
Hirnatrophie und demyelinisierende Erkrankungen des Gehirns * Gen-Panel: ID278.00 Hirnatrophie und demyelinisierende Erkrankungen des Gehirns: 55 Gene (96,0 kb) AIMP1, AIMP2, ASPA, B3GALNT2, B4GAT1, CNP, COL4A1, CRPPA, DAG1, DARS1, DEGS1, EPRS1, FAM126A, FARSA, FARSB, FKRP, FKTN, GFAP, GJC2, GMPPB, GRM7, HIKESHI, HSPD1, LARGE1, MAPT, MAT1A, MED17, MTHFS, PLP1, POLR1C, POLR3A, , POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSEN1, PYCR2, RARS1, RXYLT1, SLC25A12, SOX10, TBCD, TMEM106B, TMEM63A, TRAPPC12, TRAPPC4, TRAPPC6B, UBTF, UFM1, VPS11 Hirnatrophie: 13 Gene (24,2 kb) EXOC7, EXOC8, FARSA, FARSB, GRM7, MAPT, MED17, PSEN1, TBCD, TRAPPC4, TRAPPC6B, TRAPPC12, UBTF Hypo- und Demyelinisierung des Gehirns: 27 Gene (43,0 kb) AIMP1, AIMP2, ASPA, CNP, DARS1, DEGS1, EPRS1, FAM126A, GFAP, GJC2, HIKESHI, HSPD1, MAT1A, MTHFS, PLP1, POLR1C, POLR3A, POLR3B, PYCR2, RARS1, SLC25A12, SOX10, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Walker-Warburg-Syndrom (MDDGA): 14 Gene (23,6 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Hyperekplexie (HKPX) * Gen-Panel: ID216.00, 9 Gene (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 Wo	E
Hypomyelinisierende Leukodystrophie (HLD) * Gen-Panel: ID277.00, 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Komplexe neurologische Entwicklungsstörungen (NED) *	4 - 6 Wo	E
Gen-Panel: ID358.01, 193 Gene (496,0 kb)		
ACBD6, ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BAZ2B, BCAS3, BPTF, BRAT1, C18ORF32, CAPRIN1, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CERT1, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DPH5, DYNC1I2, EEF1D, EMC10, ESAM, EXOC2, EXOC7, EXOC8, FAM177A1, FBXW11, FDFT1, FEM1B, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, GTPBP2, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HDAC4, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCND1, KCNN2, KDM5A, KDM6B, LNPK, MADD, MAP4K4, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NAV3, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPMT1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, RNU4-2, SARS1, SEC31A, SEL1L, SETD1A, SHMT2, SHQ1, SLC4A10, SMG8, SMG9, SMPD4, SNF8, SNIP1, SPOP, SPOUT1, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, TTI1, UBE3C, UBE4A, UBR5, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZBTB11, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6		
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		
Neuronale Ceroid-Lipofuszinose (CLN) *	3 - 5 Wo	E
Gen-Panel: ID132.01, 15 Gene (20,2 kb)		
ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1		
Parkinson-Krankheit (PARK) und Parkinsonismus*	3 - 5 Wo	E
Gen-Panel: ID077.03		
Parkinson-Krankheit (PARK) und Parkinsonismus: 49 Gene (107,1 kb)		
ATP13A2, ATP1A3, ATP6AP2, C19ORF12, CHCHD2, COASY, DCTN1, DNAJC6, DNAJC12, EIF4G1, FBXO7, FTL, GBA1, GCH1, GIGYF2, GRN, HTRA2, LRRK2, MAPT, NR4A2, PARK7, PANK2, PGK1, PINK1, PLA2G6, POLG, PRKRA, PRKN, PSAP, PTPA, PTRHD1, RAB32, RAB39B, SLC18A2, SLC30A10, SLC39A14, SLC6A3, SLC9A6, SNCA, SYNJ1, TAF1, TH, TRPM7, UCHL1, UQCRC1, VPS13C, VPS35, WARS2, WDR45		
Parkinson-Krankheit (PARK), autosomal-rezessiv: 10 Gene (31,3 kb)		
ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, PTPA, SYNJ1, VPS13C		
Parkinson-Krankheit (PARK), autosomal-dominant: 11 Gene (25,5 kb)		
CHCHD2, EIF4G1, GBA1, GIGYF2, HTRA2, LRRK2, PSAP, RAB32, SNCA, UCHL1, VPS35		
Dystonie-Parkinsonismus (DYT): 12 Gene (23,6 kb)		
ATP1A3, GCH1, NR4A2, PLA2G6, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH, WARS2		
Komplexe Parkinson-Syndrome: 20 Gene (33,9 kb)		
ATP6AP2, C19ORF12, COASY, DCTN1, DNAJC12, FTL, GRN, MAPT, NR4A2, PANK2, PGK1, POLG, PTRHD1, RAB39B, SLC9A6, SLC30A10, SLC39A14, TRPM7, UQCRC1, WDR45		
Paroxysmale Dyskinesie *	3 - 5 Wo	E
Gen-Panel: ID286.00, 14 Gene (38,9 kb)		
ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24		
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		

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Spastische Paraplegie (HSP, SPG) *	4 - 6 Wo	E
Gen-Panel: ID148.05		
Spastische Paraplegie (HSP, SPG): 68 Gene (140,5 kb)		
ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, TMEM63C, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-dominant: 20 Gene (43,1 kb)		
ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-rezessiv: 51 Gene (103,6 kb)		
ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, TMEM63C, UCHL1, VPS37A, ZFYVE26		
Spastische Paraplegie (SPG), X-chromosomal: 3 Gene (6,9 kb)		
ABCD1, L1CAM, PLP1		
Spinale Muskelatrophie (SMA) *	3 - 5 Wo	E
Gen-Panel: ID152.03		
Spinale Muskelatrophie (SMA): 41 Gene (91,7 kb)		
AR, ASAH1, ASCC1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1		
Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb)		
AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB		
Distale spinale Muskelatrophie (DSMA, HMN): 26 Gene (58,1 kb)		
ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
Spinozerebelläre Ataxie, autosomal-dominant (SCA, ADCA) *	3 - 5 Wo	E
Gen-Panel: ID236.03, 27 Gene (91,6 kb)		
AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPTX1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2		
Spinozerebelläre Ataxie, autosomal-rezessiv (SCAR, SCAN) *	4 - 6 Wo	E
Gen-Panel: ID213.04, 36 Gene (117,6 kb)		
ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PNPLA6, POLG, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TWNK, UBA5, VPS13D, VPS41, VWA3B, WDR73, WWOX, XRCC1		
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		
Neuromuskuläre Erkrankungen		
Amyotrophe Lateralsklerose (ALS) *	3 - 5 Wo	E
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		
Arthrogrypose *	4 - 6 Wo	E
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		

Erkrankung/Diagnostik	Dauer	Material
Neuromuskuläre Erkrankungen		
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		
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		
Distale Arthrogrypose (DA) *	3 - 5 Wo	E
Gen-Panel: ID196.02, 11 Gene (40,5 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Distale motorische Neuronopathie (HMND, HMNR, HMNX) *	3 - 5 Wo	E
Gen-Panel: ID254.02		
Distale motorische Neuronopathie (HMND, HMNR, HMNX): 26 Gene (58,1 kb)		
ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
Distale motorische Neuronopathie, autosomal-dominant (HMND): 16 Gene (40,2 kb)		
BAG3, BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1		
Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 11 Gene (18,5 kb)		
ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, RTN2, SIGMAR1, SORD, VRK1, VWA1		

Erkrankung/Diagnostik	Dauer	Material
Neuromuskuläre Erkrankungen		
Distale Myopathie (MPD) * 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, TCAP, TIA1, TTN, VCP	4 - 6 Wo	E
Duchenne- und Becker-Muskeldystrophie (DMD, BMD) *, # Gen-Panel: ID256.00, 1 Gen (11,1 kb) DMD	2 - 4 Wo	E
Emery-Dreifuss-Muskeldystrophie (EDMD) * Gen-Panel: ID121.00, 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43	3 - 5 Wo	E
Gliedergürtelmuskeldystrophie (LGMD) * 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	4 - 6 Wo	E
Hereditäre Neuropathien (HMSN, HSAN, SMA), umfassende Diagnostik * Gen-Panel ID374.02 Hereditäre Neuropathien (HMSN, HSAN, SMA), umfassende Diagnostik: 245 Gene (598,6 kb) AAAS, AARS1, ABCA1, ABCD1, ABHD12, ACOX1, ADA2, ADCY6, ADGRG6, ADPRS, AGTPBP1, AGXT, AIFM1, AMACR, AP1S1, AP5Z1, APOA1, APTX, AR, ARHGEF10, ARL6IP1, ARSA, ASAH1, ASCC1, ATAD3A, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP7A, ATXN10, ATXN1, ATXN2, ATXN3, ATXN7, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CADM3, CAPN1, CD59, CFAP276, CHCHD10, CLP1, CNTNAP1, COA7, COQ7, COX20, COX6A1, CPOX, CTDP1, CYP27A1, CYP2U1, DARS2, DCTN1, DHH, DHTKD1, DHX9, DMXL2, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, ERCC6, ERCC8, ETFDH, EXOSC3, FA2H, FAH, FBLN5, FBXO38, FDXR, FGD4, FICD, FIG4, FLVCR1, FMR1, FXN, GALC, GAN, GARS1, GBA2, GBF1, GDAP1, GJB1, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HPDL, HSPB1, HSPB3, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, LYST, MAG, MARS1, MCM3AP, MED25, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MTTP, MYH14, NAGA, NAGLU, NARS1, NDC1, NDRG1, NDUFS6, NEFH, NEFL, NEMF, NFASC, NGF, NTRK1, NUDT2, OPA1, OPA3, PDHA1, PDK3, PDXK, PDYN, PEX10, PEX7, PHYH, PIEZO2, PIGB, PLA2G6, PLAAT3, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, PNPT1, POLG, POLR3A, POLR3B, PPOX, PRDM12, PRNP, PRPS1, PRX, PTRH2, RAB7A, REEP1, RETREG1, RTN2, SACS, SAMD9L, SARS1, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN9A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SMN1, SMN2, SORD, SOX10, SPAST, SPG11, SPG7, SPTAN1, SPTBN4, SPTLC1, SPTLC2, SURF1, SYT2, TBCE, TECPR2, TFG, TRIM2, TRIP4, TRMT5, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, UCHL1, VAPB, VCP, VPS13D, VRK1, VWA1, WARS1, WNK1, XK, XPA, YARS1, ZFYVE26	4 - 6 Wo	E
Kongenitale Myopathie (CMYP) * 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, LMOD3, MAP3K20, MEGF10, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYL2, MYOD1, MYOT, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SELENON, SPEG, STAC3, SVIL, 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	4 - 6 Wo	E
Kongenitales myasthenes Syndrom (CMS) * 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	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

Erkrankung/Diagnostik	Dauer	Material
Neuromuskuläre Erkrankungen		
Metabolische Muskelerkrankungen und Rhabdomyolyse * Gen-Panel: ID395.00, 93 Gene (205,6 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, AGK, AGL, ALDOA, AMPD1, ANO5, ATP2A2, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHKB, CNTN1, COQ4, COQ8A, CPT2, CTBP1, DMD, DTNA, DNAJB6, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FKRP, FLAD1, GAA, GBE1, GMPPB, GUK1, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LIG3, LPIN1, MGME1, MLIP, MPV17, MRM2, MYH1, OBSCN, OPA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PNPLA2, POC5, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC22A5, SLC25A10, SLC25A20, SLC25A21, SLC25A4, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TK2, TAMM41, TANGO2, TRAPPC2L, TSFM, TWNK, TYMP, YARS2	4 - 6 Wo	E
Muskeldystrophie-Dystroglykanopathie (MDDG) * Gen-Panel: ID179.00 Muskeldystrophie-Dystroglykanopathie (MDDG): 15 Gene (24,0 kb) B3GALNT2, B4GAT1, DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglykanopathie (MDDGA) mit Hirn- und Augenanomalien: 14 Gene (22,8 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglykanopathie (MDDGB) mit oder ohne intellektuelle Entwicklungsstörung: 8 Gene (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2 Muskeldystrophie-Dystroglykanopathie (MDDGC), Gliedergürtelmuskeldystrophie: 11 Gene (18,5 kb) DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, POMGNT1, POMGNT2, POMK, POMT1, POMT2	3 - 5 Wo	E
Muskelerkrankungen 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
Myopathien, umfassende Diagnostik * Gen-Panel: ID336.01 Myopathien, umfassende Diagnostik: 274 Gene (811,8 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, AGRN, ALDOA, ALG14, ALG2, AMPD1, ANO5, ASAH1, ASCC1, ATP2A1, ATP2A2, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, COQ4, COQ8A, CPT2, CRPPA, CRYAB, CTBP1, DAG1, DES, DGUOK, DMD, DMPK, DNA2, DNAJB4, DNAJB6, DNM2, DNMT3B, DTNA, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBXL4, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFPT1, GMPPB, GNE, GUK1, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HINT1, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB8, IGHMBP2, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIG3, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MAP3K20, MATR3, MEGF10, MFN2, MGME1, MICU1, MLIP, MPV17, MPZ, MRM2, MTM1, MTMR14, MTRFR, MUSK, MYBPC1, MYH1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NEB, NEFL, OBSCN, OPA1, ORAI1, PAX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PIEZO2, PLEC, PNPLA2, POC5, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RNU4-2, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SCO2, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A10, SLC25A20, SLC25A21, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC52A3, SLC5A7, SMCHD1, SMN1, SMPX, SNAP25, SPEG, SPG7, SPG11, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TAMM41, TANGO2, TCAP, TFAM, TIA1, TK2, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRAPPC2L, TRIM32, TRIP4, TRMT5, TRPV4, TSFM, TTN, TUBB3, TWNK, TYMP, UBA1, UNC45B, VAMP1, VCP, VMA21, VRK1, YARS2 Kongenitale Myopathie (CMYP): 49 Gene (268,1 kb) ACTA1, ACTN2, BAG3, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, CRYAB, DES, DNAJB4, DNM2, FLNC, FXR1, HACD1, KBTBD13, KLHL40, KLHL41, KY, LDB3, LMOD3, MAP3K20, MEGF10, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYL2, MYOD1, MYOT, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SELENON, SPEG, STAC3, SVIL, TNNC2, TNNT1, TPM2, TPM3, TTN, UNC45B Distale Myopathie (MPD): 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, TCAP, TIA1, TTN, VCP Muskeldystrophie (LGMD, EMD): 52 Gene (283,6 kb) ANO5, B3GALNT2, B4GAT1, CAPN3, CNBP, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DES, DAG1, DMD, DMPK, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, HNRNPDL, JAG2, LAMA2, LARGE1, LIMS2, LMNA, PLEC, POGLUT1, POPDC1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RXYLT1, SGCA, SGCB, SGCD, SGCG, SMCHD1, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	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

Erkrankung/Diagnostik	Dauer	Material
Neuromuskuläre Erkrankungen		
Spastische Paraplegie (HSP, SPG) *		
Gen-Panel: ID148.05		
Spastische Paraplegie (HSP, SPG): 68 Gene (140,5 kb)		
ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, TMEM63C, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-dominant: 20 Gene (43,1 kb)		
ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-rezessiv: 51 Gene (103,6 kb)		
ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, TMEM63C, UCHL1, VPS37A, ZFYVE26		
Spastische Paraplegie (SPG), X-chromosomal: 3 Gene (6,9 kb)		
ABCD1, L1CAM, PLP1		
Spinale Muskelatrophie (SMA) *		
Gen-Panel: ID152.03		
Spinale Muskelatrophie (SMA): 41 Gene (91,7 kb)		
AR, ASAH1, ASCC1, ATP7A, BAG3, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1	3 - 5 Wo	E
Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb)		
AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB		
Distale spinale Muskelatrophie (DSMA, HMN): 26 Gene (58,1 kb)		
ATP7A, BAG3, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, RTN2, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1		
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		
Zentronukleäre Myopathie (CNM) *		
Gen-Panel: ID257.00, 7 Gene (21,4 kb)	3 - 5 Wo	E
CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG		
Nierenerkrankungen		
Alport-Syndrom (ATS) *		
Gen-Panel: ID099.00, 4 Gene (21,0 kb)	3 - 5 Wo	E
COL4A3, COL4A4, COL4A5, MYH9		
Aminoazidurie *		
Gen-Panel: ID318.00		
Aminoazidurie: 13 Gene (21,1 kb)		
EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2		
Cystinurie: 2 Gene (3,5 kb)	3 - 5 Wo	E
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) *		
Gen-Panel: ID163.04, 20 Gene (42,1 kb)	3 - 5 Wo	E
ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN		
Bardet-Biedl-Syndrom (BBS) *		
Gen-Panel: ID093.02, 21 Gene (39,0 kb)	3 - 5 Wo	E
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Bartter-Syndrom (BARTS) *		
Gen-Panel: ID156.01, 8 Gene (17,7 kb)	3 - 5 Wo	E
BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3		
Branchiootorenales Syndrom (BOR) *		
Gen-Panel: ID315.00, 5 Gene (10,2 kb)	3 - 5 Wo	E
EYA1, SALL1, SIX1, SIX5, TFAP2A		
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen *		
Gen-Panel: ID706.01, 16 Gene (30,1 kb)	3 - 5 Wo	E
ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1		
Diabetes insipidus *		
Gen-Panel: ID322.00, 5 Gene (8,4 kb)	3 - 5 Wo	E
AQP2, AVP, AVPR2, SLC12A1, WFS1		
Fraser-Syndrom (FRASRS) *		
Gen-Panel: ID317.00, 3 Gene (24,8 kb)	3 - 5 Wo	E
FRAS1, FREM2, GRIP1		

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
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
Galloway-Mowat-Syndrom (GAMOS) * Gen-Panel: ID251.01, 10 Gene (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 Wo	E
Glomerulonephritis * Gen-Panel: ID103.01 Glomerulonephritis: 18 Gene (36,9 kb) C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD C3-Glomerulopathie (C3G): 3 Gene (7,2 kb) CFI, CFH, CFHR5 C1q-Mangel (C1QD): 3 Gene (2,3 kb) C1QA, C1QB, C1QC	3 - 5 Wo	E
Hyperoxalurie Gen-Panel: ID363.00 Hyperoxalurie: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb) AGXT, GRHPR, HOGA1 Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 Wo	E
Hypomagnesiämie (HOMG) * Gen-Panel ID054.02: 14 Gene (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6	3 - 5 Wo	E
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
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

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
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
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: ID361.01, 32 Gene (56,7 kb) ADCY10, AGXT, ALPL, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, FAH, FAM20A, GRHPR, HNF4A, HOGA1, KCNJ1, MAGED2, OCRL, OXGR1, PHEX, RRAGD, SLC12A1, SLC34A1, SLC34A3, SLC4A1, STRADA, VIPAS39, VPS33B	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
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
Nierensteine und Nephrokalzinose * Gen-Panel ID231.05 Nierensteine und Nephrokalzinose: 44 Gene (80,2 kb) ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, FAM20A, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, NHERF1, OCRL, OXGR1, PHEX, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, VIPAS39, VPS33B, WDR72, XDH Hyperkalziurie: 16 Gene (32,3 kb) ADCY10, BSND, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, RRAGD, SLC12A1, SLC34A1, SLC34A3 Hyperoxalurie: 5 Gene (6,3 kb) AGXT, GRHPR, HOGA1, SLC26A1, OXGR1 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Hypocitraturie: 4 Gene (11,1 kb) TP6V0A4, ATP6V1B1, SLC4A1, SLC4A4 Xanthinurie: 2 Gene (6,7 kb) MOCOS, XDH Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9	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

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
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 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
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 tubuläre Dysgenesie (RTD) * Gen-Panel: ID316.00, 4 Gene (7,7 kb) ACE, AGT, AGTR1, REN	2 - 4 Wo	E
Renotubuläres Fanconi-Syndrom (FRTS) * Gen-Panel: ID359.00, 7 Gene (10,4 kb) CTNS, EHHADH, GATM, HNF4A, NDUFAF6, SLC2A2, SLC34A1	3 - 5 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
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, THBD	3 - 5 Wo	E
Tubulointerstitielle Nierenerkrankung, autosomal-dominant (ADTKD) * Gen-Panel: ID296.00, 6 Gene (8,8 kb) DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
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
Vesikoureteraler Reflux (VUR) * Gen-Panel: ID314.00, 10 Gene (33,7 kb) DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB	3 - 5 Wo	E
Zystische Nierenerkrankungen, umfassende Diagnostik * 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	4 - 6 Wo	E
Skelett- und Knochenerkrankungen		
Adams-Oliver-Syndrom (AOS) * Gen-Panel: ID259.00, 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 Wo	E
Akrozephalosyndaktylie (ACS) * Gen-Panel: ID311.00, 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1	3 - 5 Wo	E
Arachnodaktylie * Gen-Panel: ID124.00, 13 Gene (35,9 kb) CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFBR1, TGFBR2, SMAD3	3 - 5 Wo	E
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
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 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	3 - 5 Wo	E
Distale Arthrogrypose (DA) * Gen-Panel: ID196.02, 11 Gene (40,5 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1	3 - 5 Wo	E
Fraser-Syndrom (FRASRS) * Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
Frontonasale Dysplasie (FND) * Gen-Panel ID339.00: 11 Gene (22,1 kb) ALX1, ALX3, ALX4, ANKH, EFNB1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Handfehlbildungen, umfassende Diagnostik * Gen-Panel: ID298.00 Handfehlbildungen, umfassende Diagnostik: 110 Gene (295,9 kb) ADAMTS10, ADAMTS17, AKT3, BHLHA9, BMP2, BMPR1B, C2CD3, CACNA1C, CCND2, CCNQ, CDH3, CHST11, CHSY1, CIBAR1, CKAP2L, CPLANE1, CREBBP, DACT1, DDX59, DHCR7, DHODH, DLL4, DLX5, DOCK6, EFNB1, EFTUD2, EOGT, EP300, ESCO2, EVC2, FBLN1, FBN1, FGF10, FGF16, FGF9, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GATA6, GDF5, GDF6, GJA1, GLI1, GLI2, GLI3, GRIP1, HOXA13, HOXD13, HUWE1, IFT57, IGF2, IHH, INTU, IQCE, IRF6, KIAA0753, KIAA0825, KIF7, LMBR1, LMNA, LRP4, LTBP2, MAP3K20, MECOM, MEGF8, MYCN, NAA10, NECTIN1, NECTIN4, NOG, NOTCH1, OFD1, PAX3, PDE3A, PDE4D, PIK3CA, PIK3R2, PITX1, PRKAR1A, PRMT7, PTHLH, RAB23, RBM8A, RBPJ, RECQL4, RIPK4, ROR2, RUNX2, SALL1, SALL4, SF3B4, SMO, SMOC1, SOST, TBC1D24, TBX15, TBX3, TBX5,TCTN3, TMEM107, TP63, TRPV4, TWIST1, WDPCP, WNT10B, WNT7A, YY1AP1, ZNF141 Brachydaktylie (BD), nicht-syndromal: 9 Gene (14,0 kb) BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PDE3A, PTHLH, ROR2 Polydaktylie (PAPA, PPD), nicht-syndromal: 9 Gene (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141 Syndaktylie (SDTY), nicht-syndromal: 8 Gene (18,5 kb) BHLHA9, FBLN1, GJA1, GLI3, HOXD13, LMBR1, LRP4, NECTIN4 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	4 - 6 Wo	E
Herz-Hand-Syndrom * Gen-Panel: ID165.01, 9 Gene (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 Wo	E
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

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

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
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
3M-Syndrom * Gen-Panel: ID214.00, 3 Gene (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 Wo	E
Stoffwechselerkrankungen		
Aminoazidurie * Gen-Panel: ID318.00 Aminoazidurie: 13 Gene (21,1 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2 Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Renotubuläres Fanconi-Syndrom (FRTS): 5 Gene (7,7 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC34A1	3 - 5 Wo	E
Amyloidose * Gen-Panel: ID375.00 Amyloidose: 18 Gene (25,0 kb) APP, APOA1, APOA2, APOC2, B2M, CST3, FGA, GPNMB, GSN, IL31RA, ITM2B, LYZ, MEFV, NLRP3, OSMR, PRNP, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 8 Gene (7,0 kb) APOA1, APOA2, APOC2, B2M, FGA, GSN, LYZ, TTR Primäre lokalisierte kutane Amyloidose (PLCA): 3 Gene (7,0 kb) GPNMB, OSMR, IL31RA Zerebrale Amyloidangiopathie (HCHWA): 4 Gene (4,3 kb) APP, CST3, ITM2B, PRNP	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
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		
Homocystinurie *	3 - 5 Wo	E
Gen-Panel: ID191.01, 9 Gene (15,3 kb)		
ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1		
Hyperinsulinämische Hypoglykämie (HHF) *	3 - 5 Wo	E
Gen-Panel: ID126.00, 8 Gene (16,9 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A		
Hyperkalzämie *	3 - 5 Wo	E
Gen-Panel: ID262.00, 8 Gene (14,6 kb)		
AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1		
Hyperoxalurie	3 - 5 Wo	E
Gen-Panel: ID363.00		
Hyperoxalurie: 6 Gene (8,6 kb)		
AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6		
Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb)		
AGXT, GRHPR, HOGA1		
Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb)		
OXGR1, SLC26A1, SLC26A6		
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *	3 - 5 Wo	E
Gen-Panel: ID292.00, 6 Gene (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung *	3 - 5 Wo	E
Gen-Panel: ID280.00		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung: 44 Gene (85,9 kb)		
ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4		
Glykogenspeicherkrankheit (GSD): 24 Gene (48,4 kb)		
AGL, ALDOA. ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4		
Hyperinsulinämische Hypoglykämie (HHF): 9 Gene (18,8 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A		
Hypomagnesiämie (HOMG) *	3 - 5 Wo	E
Gen-Panel ID054.02: 14 Gene (30,9 kb)		
ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6		
Hypophosphatasie, Hypophosphatämie und Rachitis *	3 - 5 Wo	E
Gen-Panel: ID269.03		
Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb)		
ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR		
Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb)		
CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3		
Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb)		
CYP2R1, CYP3A4, CYP27B1, VDR		
Hypophosphatasie (HPP): 1 Gen (1,6 kb)		
ALPL		

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Kombinierter oxidativer Phosphorylierungsmangel (COXPD) *	3 - 5 Wo	E
Gen-Panel: ID287.01, 60 Gene (80,3 kb)		
AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2		
Kongenitale Glykosylierungsstörung (CDG) *	3 - 5 Wo	E
Gen-Panel: ID035.03		
Kongenitale 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		
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		
Metabolische Muskelerkrankungen und Rhabdomyolyse *	4 - 6 Wo	E
Gen-Panel: ID395.00, 93 Gene (205,6 kb)		
ABHD5, ACAD9, ACADM, ACADS, ACADVL, AGK, AGL, ALDOA, AMPD1, ANO5, ATP2A2, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHKB, CNTN1, COQ4, COQ8A, CPT2, CTBP1, DMD, DTNA, DNAJB6, DGUOK, DYSF, ENO3, ETFA, ETFB, ETFDH, FBXL4, FDX2, FKRP, FLAD1, GAA, GBE1, GMPPB, GUK1, GYG1, GYS1, HADHA, HADHB, ISCU, LAMP2, LDHA, LIG3, LPIN1, MGME1, MLIP, MPV17, MRM2, MYH1, OBSCN, OPA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PNPLA2, POC5, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC22A5, SLC25A10, SLC25A20, SLC25A21, SLC25A4, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TK2, TAMM41, TANGO2, TRAPPC2L, TSFM, TWNK, TYMP, YARS2		
MODY-Diabetes *	3 - 5 Wo	E
Gen-Panel: ID048.01, 14 Gene (22,9 kb)		
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1		
Mukopolysaccharidose (MPS) *	3 - 5 Wo	E
Gen-Panel: ID308.00, 12 Gene (21,2 kb)		
ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A		
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		
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		

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
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		
Pyruvatdehydrogenase-Mangel (PDHD) *	3 - 5 Wo	E
Gen-Panel: ID393.00, 7 Gene (10,0 kb)		
DLAT, DLD, LIAS, PDHA1, PDHB, PDHX, PDP1		
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		
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		
Zystische Fibrose (CF) *	2 - 4 Wo	E
Gen-Panel: ID045.00, 1 Gen (4,4 kb)		
CFTR		
3-Methylglutaconazidurie (MGCA) *	3 - 5 Wo	E
Gen-Panel: ID249.01, 16 Gene (18,9 kb)		
AGK, ATPAF2, ATP5F1D, ATP5F1E, AUH, CLPB, DNAJC19, ECHS1, HTRA2, OPA3, POLG, SERAC1, SUCLA2, TAFAZZIN, TIMM50, TMEM70		

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
BRCA1- und BRCA2-assoziierte Tumordisposition *, # Gen-Panel: ID001.00, 2 Gene (15,8 kb) BRCA1, BRCA2	2 - 4 Wo	E
Chromosomen-Instabilitätssyndrome * 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	4 - 6 Wo	E
Cowden-Syndrom (CWS) * Gen-Panel: ID075.01, 8 Gene (12,8 kb) AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1	3 - 5 Wo	E
DNA-Reparatur-Defizienz-Syndrome, umfassende Diagnostik * 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	4 - 6 Wo	E
Dyskeratosis congenita (DKC) * Gen-Panel: ID347.01, 15 Gene (24,1 kb) ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53	3 - 5 Wo	E
Endometriumkarzinom * Gen-Panel: ID364.00, 12 Gene (29,1 kb) EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, STK11, TP53	3 - 5 Wo	E
Endometriumkarzinom, umfassende Diagnostik * 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	3 - 5 Wo	E
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
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
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 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

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Kolorektales Karzinom * Gen-Panel: ID006.10, 24 Gene (66,3 kb) APC, ATM, AXIN2, BAP1, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53	3 - 5 Wo	E
Konstitutionelles MMR-Defizienz-Syndrom (CMMRDS, MMRCS) * Gen-Panel: ID362.00, 5 Gene (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	2 - 4 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
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
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
Lynch-Syndrom (LYNCH, HNPCC) * Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	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
Tumorerkrankungen		
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: ID021.03, 13 Gene (41,0 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NTHL1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53	3 - 5 Wo	E
Mamma- und Ovarialkarzinom (HBOC) * Gen-Panel: ID003.05 ATM, BARD1, BRIP1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NTHL1, PALB2, PMS2, PTEN, 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
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
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: ID005.07, 16 Gene (41,6 kb) APC, AXIN2, BAP1, 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

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

Erkrankung/Diagnostik	Dauer	Material
Pränatale Diagnostik: Fetale Anomalien		
Fetale Anomalien *	4 - 6 Wo	E
Gen-Panel: ID850.00, 1223 Gene (3235,6 kb)		
AAAS, ABCA12, ABCC6, ABCC9, ABHD5, ABL1, ACAD9, ACADVL, ACAN, ACE, ACOX1, ACP5, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTG2, ACVR2B, ACY1, ADAMTS10, ADAMTS17, ADAMTS3, ADAMTSL2, ADAR, ADGRG1, ADGRG6, ADNP, ADSL, AFF4, AGK, AGL, AGPS, AHCY, AHDC1, AHI1, AKT1, AKT2, AKT3, ALDH18A1, ALDH1A3, ALDH3A2, ALDH7A1, ALDOA, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALX1, ALX3, ALX4, AMACR, AMER1, AMMECR1, AMPD2, AMT, ANAPC1, ANKH, ANKRD11, ANKS6, ANOS1, ANTXR1, ANTXR2, AP1S2, AP4E1, AR, ARCN1, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6V0A2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCL11A, BCOR, BCS1L, BFSP2, BGN, BHLHA9, BICD2, BIN1, BLM, BLTP1, BMP1, BMP2, BMP4, BMPER, BMPR1B, BNC2, BPNT2, BRAF, BRAT1, BRCA2, BRIP1, BRPF1, BSND, BTD, BUB1B, C2CD3, CA2, CA8, CACNA1C, CACNA1E, CACNA1G, CANT1, CASK, CASR, CBL, CC2D2A, CCBE1, CCDC103, CCDC39, CCDC40, CCDC8, CCDC88C, CCND2, CCNQ, CDAN1, CDC45, CDC6, CDH1, CDH3, CDK13, CDK5RAP2, CDK8, CDKL5, CDKN1C, CDON, CDT1, CELSR1, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFAP53, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD4, CHD7, CHKB, CHMP1A, CHRNA1, CHRNA3, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CILK1, CIT, CKAP2L, CLCN7, CLP1, CLPB, CNOT1, CNOT3, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COLEC10, COLEC11, COLQ, COQ4, COQ9, COX7B, CPLANE1, CPT2, CRADD, CRB2, CREB3L1, CREBBP, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CSF1R, CSNK2A1, CSPP1, CTC1, CTCF, CTNNB1, CTNND1, CTSA, CTSD, CTSK, 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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, 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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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TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRAF7, TRAIP, TRAP1, TRAPPC12, TRAPPC9, TREX1, TRIM37, TRIP11, TRIP12, TRIP4, TRMT10A, TRPS1, TRPV4, TRPV6, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TTC21B, TTC7A, TTC8, TTN, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TWIST2, TXNDC15, TXNL4A, UBA1, UBE2T, UBE3B, UBR1, UMPS, UROS, USP18, USP9X, VAMP1, VEGFC, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDPCP, WDR19, WDR26, WDR35, WDR62, WDR73, WDR81, WNT1, WNT10B, WNT5A, WNT7A, WRAP53, WT1, XRCC4, XYLT1, XYLT2, YY1, ZBTB18, ZBTB20, ZC4H2, ZEB2, ZFP57, ZIC1, ZIC2, ZIC3, ZMPSTE24, ZMYND10, ZSWIM6		

Erkrankung/Diagnostik	Dauer	Material
Präventionsdiagnostik: Carrier-Screening		
Carrier-Screening * Gen-Panel: ID350.02, 755 Gene (1852,9 kb) AAAS, ABCA12, ABCA3, ABCA4, ABCB4, ABCB11, ABCC6, ABCC8, ABCD1, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACOX1, ACSF3, ADA, ADAMTS13, ADAMTS2, ADGRG1, ADGRV1, ADSL, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMPD1, AMT, ANK1, ANO10, ANTXR2, AP1S2, AP3B1, APTX, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS9, BCHE, BCKDHA, BCKDHB, BCOR, BCS1L, BLM, BLOC1S6, BRIP1, BRWD3, BSND, BTD, BTK, CA2, CANT1, CAPN3, CASK, CASQ2, CASR, CBS, CC2D1A, CC2D2A, CCDC88C, CD19, CD3D, CD3E, CD3G, CD40LG, CDH23, CDKL5, CEP290, CEP41, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNG, CIITA, CLCN1, CLCN5, CLCN7, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COG7, COG8, 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GDAP1, GDF5, GDI1, GFM1, GJA1, GJB1, GJB2, GJB3, GJB6, GJC2, GLA, GLB1, GLDC, GLE1, GNE, GNPTAB, GNPTG, GNRHR, GNS, GP1BA, GP1BB, GP9, GPC3, GRHPR, GRIK2, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HESX1, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HMOX1, HOGA1, HPD, HPRT1, HPS1, HPS3, HPS4, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HUWE1, HSD3B2, HSD3B7, HSPG2, HYAL1, HYCC1, HYLS1, ICOS, IDS, IDUA, IFNGR1, IFNGR2, IFT80, IGBP1, IGF1, IGHMBP2, IGSF1, IL12B, IL12RB1, IL1RAPL1, IL1RN, IL2RG, IL7R, INPP5E, INS, INSR, INVS, IQCB1, ITGA6, ITGB3, ITGB4, IVD, IYD, JAK3, KCNJ1, KCNJ11, KCNQ1, KCTD7, KDM5C, KRT18, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LARGE1, LBR, LCA5, LDLR, LDLRAP1, LHCGR, LHX3, LIFR, LIG4, LIPA, LIPH, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MBTPS2, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MECP2, MED12, MED17, MEFV, MESP2, MFSD8, MGAT2, MID1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPL, MPV17, MPZ, MRE11, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTTP, MUTYH, MVK, MYD88, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFA1, NDUFAF2, NDUFAF4, NDUFAF5, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NEU1, NEUROG3, NHEJ1, NHLRC1, NHS, NLGN4X, NLRP7, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR2E3, NSUN2, NTRK1, NUP62, OAT, OCA2, OCRL, OFD1, OPA3, OPHN1, ORAI1, OSTM1, OTC, OXCT1, P3H1, PAH, PAK3, PANK2, PC, PCBD1, PCCA, PCCB, PCDH15, PCDH19, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX12, PEX13, PEX2, PEX5, PEX6, PEX7, PFKM, PHGDH, PHKB, PIGN, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POLH, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRSS12, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RDH12, RELN, REN, RFT1, RLBP1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPE65, RPGR, RPGRIP1L, RPL10, RPS6KA3, RRM2B, RS1, RTEL1, SACS, SAMD9, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO1, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SHROOM4, SIL1, SKIC3, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A1, SLC4A11, SLC5A2, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMARCAL1, SMN1, SMPD1, SNAP29, SOX3, SP110, SPR, SRD5A2, SRD5A3, ST3GAL3, ST3GAL5, STAR, STAT1, STIM1, STRA6, STX11, STXBP1, STXBP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYP, TAFAZZIN, TAT, TBCE, TCIRG1, TECPR2, TF, TFR2, TG, TGM1, TH, TIMM8A, TK2, TMC1, TMEM216, TMEM67, TNFRSF11B, TNNT1, TPO, TPP1, TRAPPC9, TREX1, TRHR, TRIM32, TRIM37, TRMU, TSEN54, TSFM, TSHB, TSHR, TSPYL1, TTN, TTPA, TUFM, TUSC3, TWNK, TYMP, TYR, TYRP1, UBA1, UBE2A, UBR1, UGT1A1, UNC13D, UPB1, UPF3B, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VDR, VIPAS39, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, VWF, WAS, WNT10A, WNT3, WNT7A, WRN, XIAP, XPA, XPC, ZDHHC9, ZFYVE26, ZIC3, ZMPSTE24, ZNF469, ZNF711	4 - 6 Wo	E
Whole Exome Sequencing (WES)		
Analysen		
WES-Solo-Exom * (Index-Patient)	1 - 3 Mo	E
WES-Trio-Exom * (Index-Patient und Eltern)	1 - 3 Mo	E
Hinweise		
***** = akkreditiertes Verfahren **HG Mannheim** und **HG München**		
# = akkreditiertes Verfahren **HG Freiburg**		
E = EDTA-Blut, 3 - 5 ml		