

SYNLAB Humangenetik

Stand: 06/2025

Next Generation Sequencing (NGS)

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Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Achromatopsie (ACHM) * Gen-Panel: ID164.02, 6 Gene (10,4 kb) ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	3 - 5 Wo	E
Albinismus, umfassende Diagnostik * Gen-Panel: ID175.05 Albinismus, umfassende Diagnostik: 33 Gene (73,8 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1 Okulokutaner Albinismus (OCA, OA): 9 Gene (13,2 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 Waardenburg-Syndrom (WS): 8 Gene (9,3 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Griscelli-Syndrom (GS): 3 Gene (8,0 kb) MLPH, MYO5A, RAB27A	3 - 5 Wo	E
Altersbedingte Makuladegeneration (AMD, ARMD) * Gen-Panel: ID186.00, 16 Gene (51,0 kb) ABCA4, APOE, ARMS2, C2, C3, C9, CFB, CFH, CFI, CST3, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, RAX2	3 - 5 Wo	E
Anophthalmie und Mikrophthalmie (MCOP) * Gen-Panel ID263.02 Anophthalmie und Mikrophthalmie (MCOP): 46 Gene (107,6 kb) ABCB6, ALDH1A3, BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3, FZD5, GDF3, GDF6, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, MITF, NAA10, NHEJ1, OTX2, PAX2, PAX6, PITX3, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, TMEM98, VAX1, VSX2, YAP1 Mikrophthalmie, isolierte Form (MCOP): 6 Gene (8,6 kb) ALDH1A3, GDF3, GDF6, MFRP, PRSS56, VSX2 Mikrophthalmie, syndromale Form (MCOPS): 13 Gene (24,9 kb) BCOR, BMP4, HCCS, HMGB3, MAB21L2, NAA10, OTX2, SOX2, RARB, RAX, STRA6, TENM3, VAX1 Mikrophthalmie und Kolobom (MCOPCB): 11 Gene (22,1 kb) ABCB6, GDF3, GDF6, FZD5, NHEJ1, PAX6, RBP4, SHH, STRA6, TENM3, VSX2 Syndrome mit Mikrophthalmie: 34 Gene (90,4 kb) BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM2, FOXE3, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MITF, NAA10, OTX2, PAX2, PITX3, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TFAP2A, TMEM98, TENM3, VAX1, YAP1	4 - 6 Wo	E
Dysgenese des vorderen Augensegmentes (ASGD) * Gen-Panel: ID182.03 Dysgenese des vorderen Augensegmentes (ASGD): 11 Gene (21,5 kb) B3GLCT, CPAMD8, CYP1B1, ELP4, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN, TRIM44 Peters-Anomalie: 7 Gene (8,8 kb) B3GLCT, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3 Axenfeld-Rieger-Anomalie: 3 Gene (3,8 kb) FOXC1, PAX6, PITX2 Aniridie (AN): 3 Gene (3,6 kb) ELP4, PAX6, TRIM44	3 - 5 Wo	E
Fraser-Syndrom (FRASRS) * Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
Fuchs-Endotheldystrophie (FECD) * Gen-Panel: ID261.00, 5 Gene (13,5 kb) AGBL1, COL8A2, SLC4A11, TCF4, ZEB1	3 - 5 Wo	E
Glaukom (GLC) * Gen-Panel: ID275.01 Glaukom (GLC): 27 Gene (53,5 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CPAMD8, CYP1B1, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, PAX6, PITX2, PITX3, PXDN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMC01, 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, TMC01, WDR36 Dysgenese des vorderen Augensegmentes (ASGD): 8 Gene (17,5 kb) CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS) * Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
Hohe Myopie (MYP) * Gen-Panel ID079.03 Hohe Myopie (MYP): 25 Gene (81,1 kb) ARR3, CACNA1F, CNGB3, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CPSF1, GPR179, GRM6, GZF1, IRX5, LOXL3, LRPAP1, LRP2, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644 Hohe Myopie, nicht-syndromale Form (MYP): 12 Gene (24,5 kb) ARR3, CNGB3, CPSF1, LOXL3, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644 Syndrome mit hoher Myopie: 14 Gene (58,7 kb) CACNA1F, COL11A1, COL2A1, COL9A1, COL9A2, COL9A3, COL18A1, GPR179, GRM6, GZF1, LRP2, IRX5, P3H2, SLITRK6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Hornhautdystrophie * Gen-Panel: ID329.01 Hornhautdystrophie: 27 Gene (68,9 kb) AGBL1, CHST6, COL8A2, COL17A1, CYP4V2, DCN, GRHL2, GSN, KERA, KRT3, KRT12, LCAT, LOXHD1, MCOLN1, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFB1, 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, TGFB1 Stromale Hornhautdystrophie: 5 Gene (11,6 kb) CHST6, DCN, PIKFYVE, TGFB1, UBIAD1 Epitheliale Hornhautdystrophie: 6 Gene (12,6 kb) COL17A1, KRT3, KRT12, MCOLN1, TACSTD2, TGFB1 Brittle-Cornea-Syndrom (BCN): 2 Gene (13,8 kb) PRDM5, ZNF468 Keratokonus (KTCN): 2 Gene (2,5 kb) TUBA3D, VSX1	3 - 5 Wo	E
Katarakt (CTRCT) * Gen-Panel: ID206.01 Katarakt (CTRCT): 37 Gene (59,7 kb) AGK, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, DNMBP, EPHA2, FOXE3, FYCO1, GCNT2, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, NHS, PITX3, SIPA1L3, SLC16A12, TDRD7, UNC45B, VIM, WFS1 Katarakt (CTRCT), autosomal-dominant: 26 Gene (29,2 kb) BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, EPHA2, GJA3, GJA8, HSF4, MAF, MIP, PITX3, SLC16A12, UNC45B, VIM, WFS1 Katarakt (CTRCT), autosomal-rezessiv: 15 Gene (30,0 kb) AGK, BFSP1, CRYAA, CRYAB, CRYBB1, CRYBB3, DNMBP, FOXE3, FYCO1, GCNT2, LEMD2, LIM2, LSS, SIPA1L3, TDRD7	3 - 5 Wo	E
Kongenitale extraokuläre Muskelfibrose (CFEOM) * Gen-Panel: ID063.00, 5 Gene (10,5 kb) COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3	3 - 5 Wo	E
Kongenitaler Nystagmus (NYS) * Gen-Panel ID331.01 Kongenitaler Nystagmus (NYS): 31 Gene (71,0 kb) AHR, ATF6, CACNA1F, CEP290, CNGA3, CNGB3, CRB1, CRX, DAGLA, DCT, FRMD7, GPR143, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, LRMDA, NMNAT1, NYX, OCA2, PAX6, ROBO1, RPE65, RPGRIP1, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1 Nystagmus, nicht-syndromal (NYS): 3 Gene (8,3 kb) FRMD7, GPR143, ROBO1 Foveahypoplasie, nicht-syndromal (FVH): 3 Gene (5,1 kb) AHR, PAX6, SLC38A8 Okulokutaner Albinismus (OCA): 7 Gene (11,1 kb) DCT, LRMDA, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Lebersche kongenitale Amaurose (LCA): 12 Gene (29,5 kb) CEP290, CRB1, CRX, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1	3 - 5 Wo	E
Kongenitale stationäre Nachtblindheit (CSNB) * Gen-Panel: ID267.00, 14 Gene (39,2 kb) CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, SAG, RHO, SLC24A1, TRPM1	3 - 5 Wo	E
Lebersche hereditäre Optikusneuropathie (LHON) # Gen-Panel: ID701.00, 17 Gene (10,5 kb) MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT-TQ, MT-TT	3 - 5 Wo	E
Lebersche kongenitale Amaurose (LCA) * Gen-Panel: ID187.01, 19 Gene (38,8 kb) AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45	3 - 5 Wo	E
Makuladystrophie (MD) * Gen-Panel: ID139.02, 18 Gene (44,6 kb) ABCA4, BEST1, CDH3, CHST6, CNGB3, CRB1, CTNNA1, ELOVL4, IMPG1, IMPG2, MAPKAPK3, MFSD8, PROM1, PRPH2, RDH12, RP1L1, SIX6, TIMP3	3 - 5 Wo	E
Morbus Stargardt (STGD) * Gen-Panel: ID102.01, 4 Gene (11,4 kb) ABCA4, ELOVL4, PROM1, PRPH2	3 - 5 Wo	E
Optikusatrophie (OPA) * Gen-Panel ID081.05 Optikusatrophie (OPA): 38 Gene (60,6 kb) ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNMT1L, 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, DNMT1L, MCAT, MECR, MIEF1, OPA1, OPA3, RTN4IP1, SSBP1, TMEM126A, YME1L1 Syndrome mit Optikusatrophie: 29 Gene (46,6 kb) ATP1A3, C19ORF12, CISD2, DNAJC30, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, TBCE, TIMM8A, UCHL1, WFS1, ZNHIT3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Okulokutaner Albinismus (OCA) * Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E
Netzhauterkrankungen, umfassende Diagnostik * Gen-Panel: ID383.01, 302 Gene (761,8 kb) ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARSG, ATF6, ATOH7, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP78, CERKL, CFAP20, CFAP410, CFAP418, CFH, CHM, CIB2, CLCC1, CLEC3B, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COQ2, COQ8B, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTNND1, CTSD, CWC27, CYP4V2, DCT, DHDDS, DHX38, DNAJC5, DRAM2, DYNC2H1, EFEMP1, ELOVL4, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FSCN2, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HGSNAT, HK1, HKDC1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF3B, KIF11, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MAN2B1, MAPKAPK3, MCOLN1, MED12, MERTK, MFRP, MFSDB, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTPP, 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, TTL5, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, VWA8, WDCPC, WDR19, WHRN, ZFYVE26, ZNF408, ZNF423, ZNF513	4 - 6 Wo	E
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB) * Gen-Panel: ID300.00 Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB): 10 Gene (17,9 kb) DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK Progressive externe Ophthalmoplegie, autosomal-dominant (PEOA): 6 Gene (12,4 kb) DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK Progressive externe Ophthalmoplegie, autosomal-rezessiv (PEOB): 5 Gene (9,2 kb) DGUOK, POLG, RNASEH1, TK2, TOP3A	3 - 5 Wo	E
Retinitis pigmentosa (RP), autosomal-dominant * Gen-Panel: ID053.03, 29 Gene (64,9 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPG1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS, VWA8	3 - 5 Wo	E
Retinitis pigmentosa (RP), autosomal-rezessiv * Gen-Panel: ID050.04, 61 Gene (162,4 kb) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513	4 - 6 Wo	E
Retinitis pigmentosa (RP), umfassende Diagnostik * Gen-Panel: ID288.02 Retinitis 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, PROX1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Usher-Syndrom (USH) * Gen-Panel: ID034.01 Usher-Syndrom (USH): 13 Gene (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN Usher-Syndrom, Typ I (USH1): 6 Gene (26,2 kb) CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G Usher-Syndrom, Typ II (USH2): 4 Gene (40,4 kb) ADGRV1, PDZD7, USH2A, WHRN Usher-Syndrom, Typ III und Typ IV (USH3, USH4): 3 Gene (3,8 kb) ARSG, CLRN1, HARS1	3 - 5 Wo	E
Vitreoretinopathie * Gen-Panel ID352.00 Vitreoretinopathie: 23 Gene (58,9 kb) ATOH7, BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, PAK2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408 Exsudative Vitreoretinopathie (EVR): 15 Gene (34,9 kb) ATOH7, CAPN5, CTNNB1, BEST1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RS1, TSPAN12, VCAN, ZNF408 Syndrome mit Vitreoretinopathie (STL, KNO): 10 Gene (27,6 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, KIF11, NDP, PAK2, RCBTB1	3 - 5 Wo	E
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPBP, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Weill-Marchesani-Syndrom (WMS) * Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD) * Gen-Panel: ID101.03, 38 Gene (94,5 kb) ABCA4, ADAM9, AIPL1, ATF6, CACNA1F, CACNA2D4, CABP4, CDH3, CDHR1, CEP78, CEP250, CFAP418, CNGA3, CNGB3, CNNM4, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RP1, RPGR, RPGRIP1, SEMA4A, TLCD3B, TTL5, UNC119	3 - 5 Wo	E
Bindegewebserkrankungen		
Bindegewebserkrankungen mit Aortenbeteiligung * Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB3R1, TGFB3R2, THSD4, TNXB, ZNF469 Thorakales Aortenaneurysma und Aortendissektion (TAA/D): 17 Gene (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB3R1, TGFB3R2, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFB3R2, TGFB3R1 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
Ehlers-Danlos-Syndrom (EDS) * Gen-Panel: ID039.05 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 8 Gene (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1 Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	3 - 5 Wo	E
Hereditäre hämorrhagische Teleangiektasie (HHT) * Gen-Panel: ID155.01, 7 Gene (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Bindegewebserkrankungen		
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2	4 - 6 Wo	E
Marfan-Syndrom (MFS) *, # Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFB1, TGFB2	2 - 4 Wo	E
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.05 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 48 Gene (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTS14, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFB1, TGFB2 Loeys-Dietz-Syndrom (LDS) und Thorakales Aortenaneurysma (AAT): 17 Gene (42,6 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Stickler-Syndrom (STL): 6 Gene (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	4 - 6 Wo	E
Osteogenesis imperfecta (OI) * Gen-Panel: ID066.02, 21 Gene (39,2 kb) ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1	3 - 5 Wo	E
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) * Gen-Panel: ID020.01 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 16 Gene (41,0 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2	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, NROB2, 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, NROB2, 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, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
Adrenogenitales Syndrom (AGS, CAH) * Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
Diabetes Insipidus * Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
Endokrine Hypertonie * Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E
Glukokortikoid-Mangel (GCCD) * Gen-Panel: ID222.00, 16 Gene (25,1 kb) AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NROB1, NR3C1, PCSK1, POMC, PROP1, STAR, TBX19, TXNRD2	3 - 5 Wo	E
Hyperaldosteronismus (HALD) * Gen-Panel: ID304.00, 6 Gene (20,6 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5	3 - 5 Wo	E
Hyperinsulinämische Hypoglykämie (HHF) * Gen-Panel: ID126.00, 8 Gene (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 Wo	E
Hyperparathyreoidismus (HRPT) * Gen-Panel: ID338.00, 10 Gene (19,2 kb) AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6	3 - 5 Wo	E
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung * Gen-Panel: ID280.00 Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung: 44 Gene (85,9 kb) ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4 Glykogenspeicherkrankheit (GSD): 24 Gene (48,4 kb) AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4 Hyperinsulinämische Hypoglykämie (HHF): 9 Gene (18,8 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A	3 - 5 Wo	E
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) * Gen-Panel: ID170.05, 40 Gene (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 Wo	E
Hypoparathyreoidismus * Gen-Panel: ID353.00, 16 Gene (24,8 kb) AIRE, CASR, CYP24A1, FAM111A, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, PTH, SLC34A1, SOX3, STX16, TBCE, TBX1	3 - 5 Wo	E
Hypophysenadenom (PITA) * Gen-Panel: ID387.01, 21 Gene (47,3 kb) AIP, CDH23, CDKN1B, DICER1, EPCAM, GNAS, GPR101, MAX, MLH1, MSH2, MSH6, MEN1, PMS2, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, USP8	3 - 5 Wo	E
Kongenitale Hypothyreose * Gen-Panel: ID369.00 Kongenitale Hypothyreose: 37 Gene (69,0 kb) CDCA8, DIO1, DUOX2, DUOX2, 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, DUOX2, IYD, SLC5A5, SLC26A4, SLC26A7, TG, TPO Kombinierter Hypophysenhormonmangel (CPHD): 8 Gene (11,9 kb) HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1	3 - 5 w	E
MODY-Diabetes * Gen-Panel: ID048.01, 14 Gene (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 Wo	E
Neuroendokrine Neoplasie * Gen-Panel: ID386.00, 19 Gene (29,2 kb) AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
Neonataler Diabetes mellitus * Panel: ID162.01 Neonataler Diabetes mellitus: 29 Gene (53,6 kb) ABCC8, BSLC2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57 Permanenter neonataler Diabetes mellitus (PNDM): 10 Gene (15,9 kb) ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57 Syndromaler neonataler Diabetes mellitus: 21 Gene (40,5 kb) BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 Wo	E
Phäochromozytom-Paragangliom-Syndrom (PPGL) * Gen-Panel: ID042.03, 14 Gene (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E
Primäre und prämatüre Ovarialinsuffizienz (POI, POF) * Gen-Panel: ID078.06 Primäre und prämatüre 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 Ovarialdysgenese (ODG): 11 Gene (19,1 kb) BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7	3 - 5 Wo	E
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) * Gen-Panel: ID250.01 Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA): 8 Gene (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 Wo	E
Schilddrüsenkarzinom * Gen-Panel: ID220.02, 26 Gene (60,5 kb) ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53	3 - 5 Wo	E
Septooptische Dysplasie * Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
Störung der Geschlechtsentwicklung (DSD) * Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NROB1, NR2F2, NR5A1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NROB1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSP01, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NROB1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
Wachstumshormonmangel (IGHD, CPHD) * Gen-Panel: ID211.02 Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNPC3, OTX2, POU1F1, PROP1, SEMA3A, SOX3 Isolierter Wachstumshormonmangel (IGHD): 5 Gene (6,4 kb) BTK, GH1, GHRHR, GHSR, SOX3 Kombinierter Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Adipositas * 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, NROB2, 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, NROB2, 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) * 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	4 - 6 Wo	E
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS) * 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, 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	4 - 6 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, TMC01, 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, TMC01, 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
Neurologische Entwicklungsstörungen (NED), umfassende Diagnostik * Gen-Panel: ID358.00, 169 Gene (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6VOA1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDF1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNPBK, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAP1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNFR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 Wo	E

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, PAPP2, 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, RFW3, RIT1, RMRP, RNP3, 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, RNP3, 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 Kardiofazlokutanes 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, ZFXH4, 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, ZFXH4, 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) * 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 Kombiniertes Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A Wachstumsstörung und Makrozephalie * Gen-Panel: ID072.03 Wachstumsstörung und Makrozephalie: 30 Gene (80,9 kb) AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12 Großwuchssyndrom und Makrozephalie: 11 Gene (33,0 kb) DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12 Noonan-Syndrom und Makrozephalie: 10 Gene (15,2 kb) BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1 3M-Syndrom * Gen-Panel: ID214.00, 3 Gene (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 Wo	E
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE) * Gen-Panel ID080.03 Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNMT1, 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, RHOTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXB1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), autosomal-dominant: 51 Gene (143,1 kb) ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CDK19, CELF2, CHD2, CUX2, CYFIP2, DNMT1, 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, RHOTB2, RNF13, SCN1A, SCN2A, SCN3A, SCN8A, SIK1, SLC1A2, SPTAN1, STXB1, 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	3 - 5 Wo	E
Benigne neonatale und infantile Krampfanfälle (BFNS, BFIS) * Gen-Panel: ID134.01, 6 Gene (19,8 kb) CHRNA2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A	3 - 5 Wo	E
Epileptische Enzephalopathien, umfassende Diagnostik * 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, ATP6VOA1, ATP6V1A, BRAT1, BSCL2, BTBD, CACNA1A, CACNA1E, CAD, CCDC88A, CDK19, CDKL5, CELF2, CHD2, CLCN4, CNPY3, CPLX1, CNTNAP2, CUX2, CYFIP2, D2HGDH, DALRD3, DENND5A, DNMT1, DNMT1L, DMXL2, DOCK7, EEF1A2, ETHE1, FGF12, FGF13, FOLR1, FOXG1, FBXO28, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GATM, 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, PCDH19, PHACTR1, PHGDH, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PPP3CA, PURA, RHOTB2, 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, STXB1, SYNGAP1, SYNJ1, SZT2, TBC1D24, 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, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNMT1, 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, RHOTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXB1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Metabolische Enzephalopathie mit Epilepsie: 29 Gene (44,8 kb) ABAT, ADSL, ALDH5A1, ALDH7A1, AMT, BTBD, D2HGDH, FOLR1, GATM, GCSH, GLDC, GPHN, IDH2, LIAS, MDH2, MOCS1, MOCS2, MTHFR, PC, PHGDH, PNPO, POLG, SLC1A2, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, TPK1	4 - 6 Wo	E

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, CHRN2, 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, RHOTB2, 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, CHRN2, 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, RHOTB2, 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, GEFSF) * Gen-Panel: ID059.03 Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFSF): 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 (GEFSF): 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, CHRN2, 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, CHRN2, 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 (GEFSF): 7 Gene (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 Wo	E
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFSF, 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, CNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETF, 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, SLC7A6OS, 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, SLC7A6OS	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Neugeborenenalter * Gen-Panel: ID135.00, 25 Gene (39,4 kb) ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter * Gen-Panel: ID171.00, 18 Gene (31,5 kb) ALDH5A1, ATP7A, BTD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Schul- und Jugendalter * Gen-Panel: ID172.00, 15 Gene (28,1 kb) ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2	3 - 5 Wo	E
Fertilitätsstörungen		
Adrenogenitales Syndrom (AGS, CAH) * Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
Azoospermie und Kryptozoospermie * Gen-Panel: ID391.00, 60 Gene (141,6 kb) ADA2, ADGRG2, AR, C14ORF39, CATIP, CATSPER1, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, FANCM, FKBP6, GCNA, HFM1, KASH5, KCTD19, KLHL10, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PMFBP1, PNLD1, 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, FSI2, IFT74, QRIH2, SPEF2, SSX1, STK33, TTC21A, TTC29, USP26, WDR19	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Fertilitätsstörungen		
Männliche Infertilität, umfassende Diagnostik * Gen-Panel ID192.06 Männliche Infertilität, umfassende Diagnostik: 126 Gene (424,9 kb) ACR, ACTL7A, ACTL9, ADA2, ADGRG2, AK7, AK9, AKAP3, AR, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRRC23, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLD1, PPP2R3C, QRIC2, RAD21L1, RBBP7, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SPO11, SSX1, STAG3, STK33, STRA8, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEK3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Störung der Spermatogenese (SPGF): 107 Gene (368,8 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CT55, CYLC1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, IFT74, IQCN, KASH5, KCNU1, KLHL10, LRRC23, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLD1, PPP2R3C, QRIC2, RBBP7, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEK3, 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
Ovarialdysgenese (ODG) * Gen-Panel: ID293.02 Ovarialdysgenese (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-Gonadendysgenese (ODG): 11 Gene (19,1 kb) BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX-Gonadendysgenese 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 prämatüre Ovarialinsuffizienz (POI, POF) * Gen-Panel: ID078.06 Primäre und prämatüre 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 Ovarialdysgenese (ODG): 11 Gene (19,1 kb) BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7	3 - 5 Wo	E
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) * Gen-Panel: ID085.03, 50 Gene (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 Wo	E
Störung der Geschlechtsentwicklung (DSD) * Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NROB1, NR2F2, NR5A1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NROB1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSP01, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NROB1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
Störung der Oozyten-, Zygoten- und Embryonen-Reifung (OZEMA) * Gen-Panel: ID239.03, 26 Gene (46,2 kb) ASTL, BTG4, CDC20, CHEK1, FBXO43, KHDC3L, KPNA7, MEI1, MOS, NLRP2, NLRP5, NLRP7, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TOP6BL, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3	3 - 5 Wo	E
Zystische Fibrose (CF) * Gen-Panel: ID045.00, 1 Gen (4,4 kb) CFTR	2 - 4 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Fertilitätsstörungen		
Weibliche Infertilität, umfassende Diagnostik * Gen-Panel ID389.00 Weibliche Infertilität, umfassende Diagnostik: 80 Gene (156,6 kb) ASTL, BMP15, BNC1, BTG4, C14ORF39, CDC20, CHEK1, CLPP, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DAP3, DIAPH2, ERAL1, ERCC6, ESR2, FANCM, FBXO43, FIGLA, FMR1, FOXL2, FSHR, GDF9, HARS2, HFM1, HROB, HSD17B4, HSD3B2, HSF2BP, INHA, KASH5, KHDC3L, KPNA7, LARS2, LHCGR, MCM8, MCM9, MEI1, MEIOB, MGA, MOS, MRPS22, MSH4, MSH5, NHEJ1, NLRP2, NLRP5, NLRP7, NOBOX, NR5A1, NUP107, PABPC1L, PADI6, PANX1, PATL2, POF1B, POR, PSMC3IP, REC114, SOHLH1, SPATA22, SPIDR, STAG3, STAR, SYCE1, SYCP2L, TLE6, TOP6BL, TP63, TRIP13, TUBB8, TWNK, WEE2, XRCC2, ZFP36L2, ZP1, ZP2, ZP3, ZSWIM7 Primäre und prämatüre 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, CIDEA, CREB3L3, CYP27A1, CYP7A1, DHCR7, DHCR24, GCKR, GK, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIMA1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPA, LPL, MTP, NPC1, NPC1L1, NPC2, PCSK9, PCYT1A, PLAAT3, PLIN1, PNPLA5, POLD1, PPARG, SAR1B, SCARB1, SMPD1, SORT1, ZMPSTE24 Fettstoffwechselstörung durch LDL-Mangel * Gen-Panel: ID094.01, 10 Gene (29,6 kb) ANGPTL3, ANGPTL4, ANGPTL8, APOB, APOE, LIMA1, MTP, NPC1L1, PCSK9, SAR1B 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 Hypertriglyceridämie * Gen-Panel: ID095.03 Hypertriglyceridämie: 32 Gene (51,1 kb) ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, 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, CIDEA, LIPE, LMNA, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24 Hypercholesterinämie * Gen-Panel: ID024.02, 13 Gene (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1	4 - 6 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, CIDEA, 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, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1 Hypertriglyceridämie: 29 Gene (51,1 kb) AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, 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, CIDEA, 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, CIDEA, 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, CIDEA, 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, SLC01B1	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
Lynch-Syndrom (LYNCH, HNPCC) * Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	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 und Polyposis * Gen-Panel ID006.09, 23 Gene (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53	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, CTSC, CTSB, GPIHBP1, LMF1, LPL, PNLI, PRSS1, SPINK1, TRPV6, UBR1	3 - 5 Wo	E
Polyposis-Syndrom (PS, FAP) * Gen-Panel: ID005.06, 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
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, SG01, 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, SG01, 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) ADAR, IFIH1, LSM11, RNASEH2A, RNASEH2B, RNASEH2C, RNU7-1, SAMHD1, TREX1	3 - 5 Wo	E
Holoprosenzephalie (HPE) * Gen-Panel: ID169.02, 17 Gene (48,8 kb) CDON, CNOT1, DLL1, DISP1, FGF8, FGFR1, GAS1, GLI2, PRRX1, PTCH1, SHH, SIX3, STAG2, STIL, TGIF1, WDR62, ZIC2	3 - 5 Wo	E
Joubert-Syndrom (JBTS) * Gen-Panel: ID028.03, 40 Gene (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 Wo	E
Komplexe kortikale Dysplasie mit weiteren Hirnfehlbildungen (CDCBM) * Gen-Panel: ID271.02, 15 Gene (52,0 kb) ADGRG1, APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2	3 - 5 Wo	E
Lissenzephalie (LIS) * Gen-Panel: ID133.00, 12 Gene (46,4 kb) ARX, CDK5, CEP85L, DCX, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A	3 - 5 Wo	E
Meckel-Syndrom (MKS) * Gen-Panel: ID032.02, 13 Gene (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 Wo	E
Neuronale Migrationsstörungen, umfassende Diagnostik * Gen-Panel: ID180.01 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, FKR, FKTN, FLNA, GMPBP, 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, FKR, FKTN, GMPBP, 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	4 - 6 Wo	E
Periventrikuläre noduläre Heterotopie (PVNH) * Gen-Panel: ID306.00, 6 Gene (26,1 kb) ARF1, ARFGEF2, ERMARD, FLNA, MAP1A, NEDD4L	3 - 5 Wo	E
Polymikrogyrie * Gen-Panel: ID176.01, 15 Gene (42,0 kb) ADGRG1, AKT3, CCND2, COL3A1, FIG4, KIFBP, OCLN, RTTN, PI4KA, PIK3CA, PIK3R2, TUBA1A, TUBA8, TUBB2B, WDR62	3 - 5 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
Schizenzephalie * Gen-Panel: ID173.00, 7 Gene (19,7 kb) COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62	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
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKR, FKTN, GMPBP, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Zerebelläre Dysgenese, X-chromosomal * Gen-Panel: ID219.00, 11 Gene (24,5 kb) ABCB7, CASK, DKC1, FMR1, L1CAM, MECP2, MID1, OFD1, OPHN1, SLC9A6, ZIC3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hämatologische Erkrankungen		
Anämien, umfassende Diagnostik * Gen-Panel: ID392.00 Anämien, umfassende Diagnostik: 188 Gene (356,9 kb) ABCB7, ABCG5, ABCG8, ADA, ADA2, ADAMTS13, ADH5, AK1, AK2, ALAD, ALAS2, ALDH2, ALDOA, AMN, ANK1, ATM, ATP11C, ATRX, BLM, BRCA1, BRCA2, BRIP1, C3, C1GALT1C1, CBLIF, CD46, CD59, CDAN1, CDIN1, CFB, CFH, CFHR1, CFHR3, CFI, COL4A1, COX4I2, CPOX, CUBN, CYB5R3, DGKE, DHFR, DKC1, DNASE2, DNAJC21, ELANE, EPAS1, EPB41, EPB42, EPO, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FOXP3, FTCD, G6PC3, G6PD, GATA1, GCLC, GLRX5, GPI, GPX1, GSR, GSS, HAMP, HBA1, HBA2, HBB, HBD, HBG1, HBG2, HEATR3, HFE, HK1, HSPA9, HSCB, IARS2, IREB2, KCNN4, KIF23, KLF1, LARS2, LCAT, LIG4, LPIN2, MAD2L2, MDM4, MMACHC, MMADHC, MPIG6B, MPL, MTHFD1, MTR, MTRR, MYSM1, NBN, NDUFB11, NHEJ1, NHLRC2, NHP2, NT5C3A, PALB2, PFKM, PGK1, PIEZO1, PKLR, PRF1, PUS1, RACGAP1, RAD51, RAD51C, RBSN, REN, RFWD3, RHAG, RMRP, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15A, RPS17, 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 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 Erythrozytose (ECYT) * Gen-Panel: ID138.02, 11 Gene (13,9 kb) BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL 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 Hämophilie * Gen-Panel: ID154.01, 5 Gene (18,9 kb) F8, F9, LMAN1, MCFD2, VWF 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
Atypisches hämolytisch-urämisches Syndrom (AHUS) * Gen-Panel: ID163.04, 20 Gene (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 Wo	E
Erythrozytose (ECYT) * Gen-Panel: ID138.02, 11 Gene (13,9 kb) BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL	3 - 5 Wo	E
Fanconi-Anämie (FANC) * Gen-Panel: ID043.02, 21 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E
Hämophilie * Gen-Panel: ID154.01, 5 Gene (18,9 kb) F8, F9, LMAN1, MCFD2, VWF	3 - 5 Wo	E
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) * Gen-Panel: ID321.01 Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML): 121 Gene (244,1 kb): ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8 Akute myeloische Leukämie (AML): 12 Gene (28,4 kb) ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53 Diamond-Blackfan-Anämie (DBA): 20 Gene (11,3 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Shwachman-Diamond-Syndrom (SDS): 4 Gene (7,2 kb) DNAJC21, EFL1, SBDS, SRP54 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Lungenfibrose und Knochenmarkinsuffizienz (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8 Dyskeratosis congenita (DKC): 13 Gene (21,8 kb) ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53 Kongenitale Neutropenie (SCN): 10 Gene (13,9 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS Fanconi-Anämie (FANC): 20 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2 Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb) MLH1, MSH2, MSH6, PMS2	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Hämatologische Erkrankungen		
Knochenmarkinsuffizienz (BMF) * Gen-Panel: ID357.00 Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb): ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTE11, SRP72, TERC, TERT, TP53, ZCCHC8 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTE11, TERC, TERT, ZCCHC8	3 - 5 Wo	E
Neutropenie * Gen-Panel: ID189.02 Neutropenie: 30 Gene (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndrome mit Neutropenie: 21 Gene (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, USB1, VPS13B, WDR1	3 - 5 Wo	E
Porphyrrie * 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
Thrombozytopathie (BDPLT, HPS) * 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	3 - 5 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
Haut- und Zahnerkrankungen		
Adams-Oliver-Syndrom (AOS) * Gen-Panel: ID259.00, 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 Wo	E
Albinismus, umfassende Diagnostik * Gen-Panel: ID175.05 Albinismus, umfassende Diagnostik: 33 Gene (73,8 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1 Okulokutaner Albinismus (OCA, OA): 9 Gene (13,2 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 Waardenburg-Syndrom (WS): 8 Gene (9,3 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Griscelli-Syndrom (GS): 3 Gene (8,0 kb) MLPH, MYO5A, RAB27A	3 - 5 Wo	E
Amelogenesis imperfecta (AI) * Gen-Panel: ID232.01, 17 Gene (29,4 kb) ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAF, RELT, SLC24A4, WDR72	3 - 5 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
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 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
Ektodermale Dysplasie (ECTD) * 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	3 - 5 Wo	E
Ektodermale Dysplasien, umfassende Diagnostik * 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	4 - 6 Wo	E
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

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Gorlin-Syndrom (BCNS) und ähnliche Krankheitsbilder * Gen-Panel: ID174.02, 8 Gene (25,8 kb) BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 Wo	E
Hereditäres Angioödem (HAE) * Gen-Panel: ID345.00, 7 Gene (16,4 kb) ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS) * Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
Hydrops fetalis * Gen-Panel: ID370.00, 148 Gene (404,7 kb) ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNG, COL2A1, CTSA, DHCR24, DHCR7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHBP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUFB10, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	3 - 5 w	E
Hypotrichose, nicht-syndromale Form (HYPT) * Gen-Panel: ID146.01, 12 Gene (20,1 kb) APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE	3 - 5 Wo	E
Ichthyose * Gen-Panel: ID217.01 Ichthyose: 38 Gene (73,0 kb) ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CLDN10, CSTA, CYP4F22, ELOVL4, ERCC2, FLG, GJB2, KRT1, KRT10, KRT2, LIPN, LORICRIN, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, TGM1 Kongenitale Ichthyose, autosomal-rezessiv (ARCI): 14 Gene (28,7 kb) ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1 Ichthyose, autosomal-dominant und X-chromosomal: 7 Gene (21,3 kb) ASPRV1, FLG, GJB2, KRT1, KRT2, KRT10, STS	3 - 5 Wo	E
Kutanes malignes Melanom (CMM) * Gen-Panel: ID193.01, 12 Gene (26,4 kb) BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3	3 - 5 Wo	E
Multiples Pterygium-Syndrom * Gen-Panel: ID158.01, 8 Gene (16,8 kb) CHRNA1, CHRN1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	3 - 5 Wo	E
Neurofibromatose (NF) *, # Gen-Panel: ID210.00, 3 Gene (11,6 kb) NF1, NF2, SPRED1	3 - 5 Wo	E
Okulokutaner Albinismus (OCA) * Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E
Orofaziodigitales Syndrom (OFD) * Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E
Pachyonychia congenita (PC) * Gen-Panel: ID120.01, 13 Gene (30,1 kb) AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3	3 - 5 Wo	E
Palmoplantarkeratose (PPK) * Gen-Panel: ID323.00 Palmoplantarkeratose (PPK): 33 Gene (58,2 kb) AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB3, GJB4, JUP, KDSR, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT10, KRT14, KRT16, KRT17, KRT83, LORICRIN, MBTPS2, PERP, RHBD2, SERPINB7, SLURP1, SMARCA1, 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, RHBD2, SLURP1, SMARCA1, TAT, TRPV3, WNT10A Erythrokeratoderma variabilis et progressiva (EKVP): 7 Gene (9,5 kb) GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4 Pachyonychia congenita (PC): 4 Gene (6,1 kb) KRT6A, KRT6B, KRT16, KRT17	3 - 5 Wo	E
Peeling-Skin-Syndrom (PSS) * Gen-Panel: ID309.00, 6 Gene (13,6 kb) CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5	3 - 5 Wo	E
Pierre-Robin-Sequenz * Gen-Panel: ID294.00, 33 Gene (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Primäres Lymphödem (LMPHM) * Gen-Panel: ID372.00, 21 Gene (69,4 kb) ADAMTS3, ANGPT2, CALCRL, CCB1, CELSR1, EPHB4, ERG, FAT4, FLT4, FOXC2, GATA2, GJA1, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, THSD1, TIE1, VEGFC	3 - 5 Wo	E
Progerie und progeroide Syndrome * Gen-Panel: ID147.01, 25 Gene (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 Wo	E
Selektive Zahn-Agenesie (STHAG) * Gen-Panel: ID151.02, 16 Gene (26,9 kb) EDA, EDAR, EDARADD, GREM2, IRF6, KDF1, KREMEN1, LRP6, LTBP3, MSX1, PAX9, PTH1R, TP63, TSPEAR, WNT10A, WNT10B	3 - 5 Wo	E
Systemerkrankungen mit multiplen Café-au-lait-Flecken * Gen-Panel ID351.00 Systemerkrankungen mit multiplen Café-au-lait-Flecken: 9 Gene (27,6 kb) BRAF, MLH1, MSH2, MSH6, NF1, PMS2, PTPN11, RAF1, SPRED1 Konstitutionelle Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb) MLH1, MSH2, MSH6, PMS2 Neurofibromatose (NF1, NFLS): 2 Gene (9,8 kb) NF1, SPRED1 LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 Wo	E
Tuberöse Sklerose (TSC) * Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
Xeroderma pigmentosum (XP) * Gen-Panel: ID282.00, 10 Gene (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 Wo	E
Herz- und Gefäßerkrankungen		
Alagille-Syndrom (ALGS) * Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC) * Gen-Panel: ID010.03, 27 Gene (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNT3, TNNT2, TPM1, TTN	4 - 6 Wo	E
Atriumseptumdefekt und Ventrikelseptumdefekt (ASD, VSD, AVSD) * Gen-Panel: ID143.00, 12 Gene (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1	3 - 5 Wo	E
Bikuspide Aortenklappe (AOVD) * Gen-Panel: ID301.00, 6 Gene (16,7 kb) GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2	3 - 5 Wo	E
Bindegewebserkrankungen mit Aortenbeteiligung * Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCG6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, TNXB, ZNF469 Thorakales Aortenaneurysma und Aortendissektion (TAA/D): 17 Gene (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFB2, TGFB1 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
Brugada-Syndrom (BRGDA) * Gen-Panel: ID014.02, 23 Gene (56,4 kb) ABCG9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4	3 - 5 Wo	E
CADASIL und CARASIL *, # Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
CHARGE-Syndrom * Gen-Panel: ID307.00, 3 Gene (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 Wo	E
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
Dilatative Kardiomyopathie (CMD, DCM) * Gen-Panel: ID008.05, 68 Gene (301,9 kb) ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1	4 - 6 Wo	E
Ehlers-Danlos-Syndrom (EDS) * Gen-Panel: ID039.05 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 8 Gene (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1 Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	3 - 5 Wo	E
Endokrine Hypertonie * Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E
Fallot-Tetralogie (TOF) * Gen-Panel: ID144.01, 12 Gene (23,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2	3 - 5 Wo	E
Familiäres Vorhofflimmern (ATFB) * Gen-Panel: ID016.02, 17 Gene (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A	3 - 5 Wo	E
Frühes Repolarisationssyndrom (ERS) * Gen-Panel: ID330.00, 12 Gene (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A	3 - 5 Wo	E
Isolierte kongenitale Herzfehler * Gen-Panel: ID017.04 Isolierte kongenitale Herzfehler: 43 Gene (103,7 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Atrium-/Ventrikelseptumdefekt (ASD, VSD): 12 Gene (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NR2F2, NKX2-5, TBX20, TLL1 Fallot-Tetralogie (TOF): 12 Gene (23,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2 Viszerale Heterotaxie (HTX): 13 Gene (26,2 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, MMP21, MNS1, NODAL, PKD1L1, ZIC3 Konotrunkale Herzfehlbildung (CTHM): 12 Gene 24,8 (kb) FLT4, FOXH1, CFC1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3 Aortenklappenerkrankung (AOVD): 7 Gene (19,1 kb) ELN, GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2 Hypoplastisches Linksherzsyndrom (HLHS): 5 Gene (5,2 kb) GDF1, GJA1, HAND1, NKX2-5, NR2F2 Kongenitale multiple Herzfehlbildung (CHTD): 7 Gene (14,4 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, TAB2, ZIC3 Herzklappendysplasie (CVDP): 3 Gene (14,8 kb) ADAMTS19, FLNA, PLD1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Hereditäre hämorrhagische Teleangiectasie (HHT) * Gen-Panel: ID155.01, 7 Gene (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 Wo	E
Herz-Hand-Syndrom * Gen-Panel: ID165.01, 9 Gene (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 Wo	E
Hypertrophe Kardiomyopathie (CMH, HCM) * Gen-Panel: ID007.05, 56 Gene (253,8 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, LAMP2, KRAS, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL	4 - 6 Wo	E
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, GPD1, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2V, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4 Frühes Repolarisationssyndrom (ERS): 12 Gene (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A Familiäres Vorhofflimmern (ATFB): 17 Gene (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A Short-QT-Syndrom (SQT): 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCL4A3 Sick-Sinus-Syndrom (SSS): 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN Arrhythmogene rechtsventrikuläre Dysplasie (ARVD): 15 Gene (150,6 kb) CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, PLN, LMNA, PKP2, PRKAG2, RYR2, TGFB3, TMEM43, TTN	4 - 6 Wo	E
Kardiomyopathien, umfassende Diagnostik * Gen-Panel: ID027.05 Kardiomyopathien, umfassende Diagnostik: 154 Gene (487,0 kb) ABCC9, ACTA1, ACTC1, ACTN2, ALPK3, ANKRD1, APOA1, B2M, BAG3, BAG5, BRAF, CACNA1C, CACNB2, CALR3, CAP2, CASQ2, CAV3, CDH2, COA5, COA6, CORIN, COX15, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DMPK, DNJC19, 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, SCD, 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 Nichtdilatierende linksventrikuläre Kardiomyopathie (NDLVC, LVNC): 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) * Gen-Panel: ID012.03, 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN	3 - 5 Wo	E
Konotrunkale Herzfehlbildung (CTHM) * Gen-Panel: ID160.01, 12 Gene (24,8 kb) CFC1, FLT4, FOXH1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Kongenitale Herzfehler, umfassende Diagnostik * Gen-Panel: ID019.02 Kongenitale Herzfehler, umfassende Diagnostik: 149 Gene (472,9 kb) ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR2B, ADAMTS10, ADAMTS17, ADAMTS19, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CFAP45, CFAP52, CFAP53, CFC1, CHD4, CHD7, CIROP, CITED2, CREBBP, CRELD1, DHCR7, DLL4, DNAAF1, DNAH5, DNAH9, DNAH11, DOCK6, DPYSL5, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FLT4, FOXC1, FOXF1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAAO, HAND1, HOXA1, HRAS, ISL1, JAG1, KDM6A, KMT2D, KRAS, KYN1, 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, TBX2, TBX3, TBX5, TFAP2B, TGDS, TGFB1, TGFB2, TKT, TLL1, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WPCP, 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, KYN1, 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, TGFB1, TGFB2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WPCP, YY1AP1, ZEB2, ZIC3	4 - 6 Wo	E
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2	4 - 6 Wo	E
Long-QT-Syndrom (LQT) * Gen-Panel: ID013.01, 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN	3 - 5 Wo	E
Marfan-Syndrom (MFS) *, # Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFB1, TGFB2	2 - 4 Wo	E
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.05 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 48 Gene (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFB1, TGFB2 Loeys-Dietz-Syndrom (LDS) und Thorakales Aortenaneurysma (AAT): 17 Gene (42,6 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Stickler-Syndrom (STL): 6 Gene (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	4 - 6 Wo	E
Multipler kongenitaler Herzdefekt (CHTD) * Gen-Panel: ID302.01, 9 Gene (21,5 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, PLXND1, SMAD2, TAB2, ZIC3	3 - 5 Wo	E
Muskelerkrankungen mit Herzbeteiligung * Gen-Panel: ID123.03 Muskelerkrankungen mit Herzbeteiligung: 19 Gene (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss-Muskeldystrophie (EMDM): 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker-Muskeldystrophie (DMD, BMD): 1 Gen (11,1 kb) DMD	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Nichtdilatierende linksventrikuläre Kardiomyopathie (NDLVC, LVNC) * Gen-Panel: ID011.04, 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
Noonan-Syndrom (NS) * Gen-Panel: ID023.06, 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 Wo	E
Plötzlicher Herztod * Gen-Panel: ID349.01 Plötzlicher Herztod: 127 Gene (393,9 kb) ABCC9, ACTC1, ACTN2, AKAP9, ALG10B, ALPK3, ANK2, ANKRD1, BAG3, BAG5, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAP2, CASQ2, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGF12, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GJA5, GLA, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLHL24, LAMA4, LAMP2, LDB3, LEMD2, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, NPPA, NUP155, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RANGRF, RBM20, RPL3L, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SEMA3A, SGCD, SLC4A3, SLMAP, SNTA1, TAFAZZIN, TBX5, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSPYL1, TTN, TTR, VCL, VEZF1 Kardiale Arrhythmien (BRGDA, LQT) und plötzlicher Herztod: 74 Gene (281,6 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FGF12, GJA5, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYBPC3, MYH6, MYL4, NKX2-5, NPPA, NUP155, PPA2, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TSPYL1, TTN Kardiomyopathien (HCM, DCM) und plötzlicher Herztod: 71 Gene (261,3 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, BAG5, CAP2, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSG2, DSP, EMD, EYA4, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GLA, JPH2, JUP, KLHL24, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, TAFAZZIN, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL, VEZF1	4 - 6 Wo	E
Pulmonale Hypertonie (PAH, PPH) * Gen-Panel: ID281.01, 23 Gene (49,9 kb) ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4	3 - 5 Wo	E
RASopathien * Gen-Panel: ID015.05 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan-Syndrom (NS): 15 Gene (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 Wo	E
Restriktive Kardiomyopathie (RCM) * Gen-Panel: ID105.03, 15 Gene (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR	4 - 6 Wo	E
Syndromale kongenitale Herzfehler * Gen-Panel ID252.02 Synromale kongenitale Herzfehler: 109 Gene (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFB1, TGFB2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3 Noonan-Syndrom (NS): 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 Ritscher-Schinzel-Syndrom (RTSC): 4 Gene (10,0 kb) DPYSL5, CCDC22, VPS35L, WASHC5 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Adams-Oliver-Syndrom (AOS): 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ Kabuki-Syndrom (KABUK): 2 Gene (20,8 kb) KDM6A, KMT2D Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFB1, TGFB2 CHARGE-Syndrom: 2 Gene (11,3 kb) CHD7, SEMA3E VCRL-Syndrom: 3 Gene (4,4 kb) HAAO, KYNU, NADSYN1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Short-QT-Syndrom (SQT) * Gen-Panel: ID233.01, 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3	3 - 5 Wo	E
Sick-Sinus-Syndrom (SSS) * Gen-Panel: ID107.01, 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A	3 - 5 Wo	E
Speicherkrankheiten mit Herzbeteiligung * Gen-Panel: ID149.03 Speicherkrankheiten mit Herzbeteiligung: 16 Gene (20,7 kb) APOA1, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Kardiale Glykogenspeicherkrankheit (GSD): 3 Gene (5,8 kb) GAA, LAMP2, PRKAG2 Hämochromatose (HFE): 6 Gene (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Amyloidose (AMYLD): 6 Gene (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) * Gen-Panel: ID020.01 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 16 Gene (41,0 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Nicht-syndromales thorakales Aortenaneurysma (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2	3 - 5 Wo	E
Viszerale Heterotaxie (HTX) * Gen-Panel: ID145.01, 18 Gene (72,4 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, DNAF1, 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, TGFB1, TGFB2, 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, TGFB1, TGFB2 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, X-chromosomal (DFNX) * Gen-Panel: ID290.01, 8 Gene (17,1 kb) AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX, TIMM8A	3 - 5 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

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, 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
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) * Gen-Panel: ID382.00 Antikörpermangelsyndrome (CVID, AGM): 38 Gene (70,7 kb) AICDA, BLNK, BTK, CARD11, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, CTNNB1, CXCR4, FNIP1, ICOS, IGLL1, IKZF1, IL21, IRF2BP2, LRBA, LRRC8A, MS4A1, NFKB1, NFKB2, OAS1, PIK3CD, PIK3R1, PLCG2, RAC2, SEC61A1, SH3KBP1, SLC39A7, SPI1, TCF3, TNFRSF13B, TNFRSF13C, TOP2B, UNG Allgemeiner variabler Immundefekt (CVID): 14 Gene (28,0 kb) CD19, CD81, CR2, ICOS, IKZF1, IL21, IRF2BP2, LRBA, MS4A1, NFKB1, NFKB2, SEC61A1, TNFRSF13B, TNFRSF13C Agammaglobulinämie (AGM): 11 Gene (16,2 kb) BLNK, BTK, CD79A, CD79B, IGLL1, LRRC8A, PIK3R1, SH3KBP1, SLC39A7, SPI1, TCF3 Immundefekt mit Hyper-IgM (HIGM): 4 Gene (3,2 kb) AICDA, CD40, CD40LG, UNG	3 - 5 Wo	E
Atypisches hämolytisch-urämisches Syndrom (AHUS) * Gen-Panel: ID163.04, 20 Gene (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 Wo	E
Autoinflammatorische Erkrankungen, umfassende Diagnostik * Gen-Panel: ID087.05 Autoinflammatorische Syndrome, umfassende Diagnostik: 51 Gene (99,3 kb) ADA2, ALPK1, ARPC1B, ARPC5, CARD14, DNASE2, DOCK11, DPP9, ELANE, ELF4, HCK, IKBKG, IL1RN, IL36RN, JAK1, LPIN2, LYN, MEFV, MVK, NCKAP1L, NFKB1, NLRCA, 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, NLRCA, NLRP12, NLRP3, NOD2, OTULIN, PSMB8, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 Wo	E
Chronische Granulomatose (CGD) * Gen-Panel: ID379.00, 8 Gene (8,8 kb) CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2	3 - 5 Wo	E
Hydrops fetalis * Gen-Panel: ID370.00, 148 Gen (404,7 kb) ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNG, COL2A1, CTSA, DHCR24, DHCR7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHBP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUF810, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	4 - 6 Wo	E
Hyper-IgE-Syndrom mit rekurrenten Infektionen (HIES) * Gen-Panel: ID240.01, 10 Gene (29,5 kb) DOCK8, DSG1, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TYK2, ZNF341	3 - 5 Wo	E
Knochenmarkinsuffizienz (BMF) * Gen-Panel: ID357.00 Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb): ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTE1, SRP72, TERC, TERT, TP53, ZCCHC8 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTE1, TERC, TERT, ZCCHC8	3 - 5 Wo	E
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, CTNNB1, CTSP1, 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, NLRCA, 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, RELB, RFX5, RFXANK, RFXAP, RHBDF2, RHOH, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPSA, RTE1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCGN, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLC9A3, SLC02A1, SMARCA1, 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, TFR2, TGFB1, TGFB1, TGFB2, TICAM1, TIN2, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF14, 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, ZNF51	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Immunologische Erkrankungen		
Periodische Fiebersyndrome * Gen-Panel: ID088.04, 12 Gene (26,3 kb) ELANE, MEV, MVK, NLR4, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 Wo	E
Neutropenie * Gen-Panel: ID189.02 Neutropenie: 30 Gene (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndrome mit Neutropenie: 21 Gene (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, USB1, VPS13B, WDR1	3 - 5 Wo	E
Schwerer kombinierter Immundefekt (SCID) * Gen-Panel: ID381.00, 21 Gene (47,6 kb) ADA, AK2, BCL11B, CD3E, CD3D, CIITA, CORO1A, DCLRE1C, FOXP1, IL2RG, IL7R, LAT, LIG4, JAK3, NHEJ1, POLD3, PRKDC, PTPRC, RAC2, RAG1, RAG2	3 - 5 Wo	E
Thrombotische Mikroangiopathie (TMA) * Gen-Panel: ID707.00, 23 Gene (44,7 kb) ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG,	3 - 5 Wo	E
Lungen- und Respiratorische Erkrankungen		
Chronische Granulomatose (CGD) * Gen-Panel: ID379.00, 8 Gene (8,8 kb) CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2	3 - 5 Wo	E
Interstitielle Lungenerkrankung und Lungenfibrose * Gen-Panel: ID341.01 Interstitielle Lungenerkrankung und Lungenfibrose: 40 Gene (80,9 kb) ABCA3, ACD, AP3B1, AP3D1, COPA, CSF2RA, CSF2RB, DCLRE1B, DKC1, FAM111B, FARSA, FARSB, FLNA, FOXF1, GBA1, HPS1, HPS4, ITGA3, MARS1, NAF1, NHP2, NKX2-1, NOP10, OAS1, PARN, RPA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STING1, TERC, TERT, TINF2, WRAP53, ZCCHC8 Pulmonale Alveolarproteinoase (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, GPM2, HYL1, 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, GPM2, HYL1, 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, TMC01, 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, TMC01, 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, CTNBN1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNP2, 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, CTNBN1, 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, HNRNP2, 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, ARFGF2, ASPM, ATR, BLM, CARS1, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CENPT, CEP135, CEP152, CEP63, CIT, CKAP2L, COPB1, COPB2, CRIPT, CTNBN1, 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, MSMD1, 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, RITN, 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 Zerebrokulo-fazioskelettales 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, CTNBN1, 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, TMC01, 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, TMC01, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9	4 - 6 Wo	E
Pontozebelläre Hypoplasie (PCH) * Gen-Panel: ID071.02, 26 Gene (60,5 kb) AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34 TSEN54, VPS51, VPS53, VRK1	3 - 5 Wo	E
Primäre Mikrozephalie, autosomal-rezessiv (MCPH) * Gen-Panel: ID031.02, 25 Gene (107,1 kb) ANKLE2, ASPM, CDK6, CIT, CDK5RAP2, CENPE, CENPJ, CEP135, CEP152, COPB2, KIF14, KNL1, MAP11, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PHC1, SASS6, STIL, WDFY3, WDR62, ZNF335	4 - 6 Wo	E
Seckel-Syndrom (SCKL) * Gen-Panel: ID113.00, 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP	3 - 5 Wo	E
Wachstumsstörung und Makrozephalie * Gen-Panel: ID072.03 Wachstumsstörung und Makrozephalie: 30 Gene (80,9 kb) AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12 Großwuchssyndrom und Makrozephalie: 11 Gene (33,0 kb) DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12 Noonan-Syndrom und Makrozephalie: 10 Gene (15,2 kb) BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1	3 - 5 Wo	E
Mitochondriopathien		
Defizienz der mitochondrialen Komplexe I bis V (MCDN) * Panel: ID074.03 Defizienz der mitochondrialen Komplexe I bis V (MCDN): 83 Gene (60,7 kb) ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX5A, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, CYC1, DNAJC30, FOXRED1, LRPPRC, LYRM7*, MTFMT, NDUFA1, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, PET100, PET117, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SURF1, TACO1, TIMMDC1, TMEM70, TMEM126B, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ Mitochondrialer Komplex-I-Mangel, kernkodierter Typ (MC1DN): 39 Gene (29,3 kb) ACAD9, DNAJC30, FOXRED1, MTFMT, NDUFA1, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, TIMMDC1, TMEM126B Mitochondrialer Komplex-II-Mangel, kernkodierter Typ (MC2DN): 4 Gene (3,7 kb) SDHA, SDHAF, SDHB, SDHD Mitochondrialer Komplex-III-Mangel, kernkodierter Typ (MC3DN): 10 Gene (7,1 kb) BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1 Mitochondrialer Komplex-IV-Mangel, kernkodierter Typ (MC4DN): 23 Gene (15,8 kb) COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, COX5A, LRPPRC, NDUFA4, PET100, PET117, SCO1, SCO2, SURF1, TACO1 Mitochondrialer Komplex-V-(ATP-Synthase-)Mangel, kernkodierter Typ (MC5DN): 7 Gene (4,8 kb) ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, TMEM70	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Mitochondriopathien		
Coenzym-Q10-Mangel (COQ10D) * 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
Kombinierter Defekt der oxidativen Phosphorylierung (COXPD) * Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIEP, 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, TMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 Wo	E
Lebersche hereditäre Optikusneuropathie (LHON) # Gen-Panel: ID701.00, 17 Gene (10,5 kb) MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT-TQ, MT-TT	3 - 5 Wo	E
MELAS-Syndrom # Gen-Panel: ID700.01, 22 Gene (9,2 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-RNR2, MT-TC, MT-TE, MT-TF, MT-TH, MT-TK, MT-TL1, MT-TL2, MT-TQ, MT-TS1, MT-TS2, MT-TV, MT-TW	3 - 5 Wo	E
Mitochondriales DNA-Depletionssyndrom (MTDPS) * Gen-Panel: ID324.00, 19 Gene (25,6 kb) AGK, DGUOK, FBXL4, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP	3 - 5 Wo	E
Mitochondriale Myopathie / Enzephalopathie # Gen-Panel: ID702.00, 27 Gene (7,0 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND2, MT-ND5, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TW	3 - 5 Wo	E
Mitochondriengenom # Gen-Panel: ID703.00, 37 Gene (11,4 kb) MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY	3 - 5 Wo	E
Nukleär-kodierte mitochondriale Erkrankungen * Gen-Panel: ID704.01, 312 Gene (406,1 kb) AARS2, ABCB7, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, AC02, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH4A1, ALDH6A1, AMACR, AMT, ANO10, APTX, ATL1, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATPAF2, AUH, BCAT2, BCKDHA, BCKDHB, BCS1L, BOLA3, BTBD, 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, DNML1, EARS2, ELAC2, ERCC6, 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, MIEP, 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, TFAZZIN, TARS2, TFAM, TIMM22, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UNG, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, VARS2, WFS1, WWOX, XPNPEP3, YARS2, YWHAE	4 - 6 Wo	E
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB) * Gen-Panel: ID300.00 Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB): 10 Gene (17,9 kb) DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK Progressive externe Ophthalmoplegie, autosomal-dominant (PEOA): 6 Gene (12,4 kb) DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK Progressive externe Ophthalmoplegie, autosomal-rezessiv (PEOB): 5 Gene (9,2 kb) DGUOK, POLG, RNASEH1, TK2, TOP3A	3 - 5 Wo	E
Pyruvatdehydrogenase-Mangel (PDHD) Gen-Panel: ID393.00, 7 Gene (10,0 kb) DLAT, DLD, LIAS, PDHA1, PDHB, PDHX, PDP1	3 - 5 Wo	E

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, TNNT2, 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 Defekt der oxidativen Phosphorylierung (COXPD) * Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPPE, 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, PTCO3, 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 Glykosylierungsstörung, 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, TGFB1, TGFB2	2 - 4 Wo	E
Noonan-Syndrom (NS) * Gen-Panel: ID023.06, 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, NRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 Wo	E
Orofaziodigitales Syndrom (OFD) * Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E
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 Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3 Tuberöse Sklerose (TSC) * Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2 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 Weill-Marchesani-Syndrom (WMS) * Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 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
	3 - 5 Wo	E
	2 - 4 Wo	E
	3 - 5 Wo	E
	3 - 5 Wo	E
	3 - 5 Wo	E
Muskelerkrankungen		
Arthrogrypose * Gen-Panel: ID200.01 Arthrogrypose: 61 Gene (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNA1, 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, RAPSIN, 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 Kontrakturensyndrom (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, RAPSIN	4 - 6 Wo	E
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik * Gen-Panel: ID312.02 Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik: 87 Gene (217,3 kb) AARS1, AIFM1, ARHGAP10, 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 Déjérine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46 Distale motorische Neuropathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN) * 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 Optikusatrophy (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46	4 - 6 Wo	E
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN) * 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	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
Distale hereditäre motorische Neuronopathie (HMND, HMNR) * Gen-Panel: ID254.01 Distale hereditäre motorische Neuronopathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Distale motorische Neuronopathie, autosomal-dominant (HMND): 15 Gene (38,4 kb) BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1 Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 10 Gene (14,4 kb) ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1	3 - 5 Wo	E
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, TIA1, TCAP, TTN, VCP	4 - 6 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 (LGMD): 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

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
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
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, LMOD2, MAP3K20, MEGF10, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYL2, MYOD1, MYOT, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SVIL, TPM2, SELENON, SPEG, STAC3, TNNC2, TNNT1, TPM2, TPM3, TTN, UNC45B Central-Core-Myopathie (CCD): 1 Gen (15,1 kb) RYR1 Nemalin-Myopathie (NEM): 11 Gene (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3 Zentronukleäre Myopathie (CNM): 7 Gene (20,3 kb) BIN1, CCDC78, DNM2, MAP3K20, MTM1, MTMR14, SPEG Myofibrilläre Myopathie (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYOT, PYROXD1, TTN	4 - 6 Wo	E
Letales kongenitales Kontrakturensyndrom (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
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
Muskeldystrophie, Typ Duchenne oder Becker (DMD, BMD) * Gen-Panel: ID256.00, 1 Gen (11,1 kb) DMD	2 - 4 Wo	E
Muskelerkrankungen, umfassende Diagnostik * Gen-Panel: ID336.00 Muskelerkrankungen, umfassende Diagnostik: 246 Gene (749,8 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, AGRN, ALDOA, ALG14, ALG2, AMPD1, ANO5, ASAH1, ASCC1, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNA1, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNA2, DNAJB6, DNM2, DNMT3B, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFPT1, GMPPB, GNE, GYG1, GYS1, HADH, HADHA, HADHB, HINT1, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB8, IGHMBP2, INPP5K, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIG3, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MAP3K20, MATR3, MEGF10, MFN2, MGME1, MICU1, MPV17, MPZ, MTM1, MTMR14, MTRFR, MUSK, MYBPC1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, NEFL, OPA1, ORAI1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, POGlut1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SCO2, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC25A3, SLC5A7, SMCHD1, SMN1, SNAP25, SPEG, SPG7, SPG11, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TCAP, TFAM, TIA1, TK2, TMEM43, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TRMT5, TRPV4, TSFM, TTN, TUBB3, TWNK, TYMP, UBA1, UNC45B, VAMP1, VCP, VMA21, VRK1, YARS2 Kongenitale, distale und metabolische Myopathien: 110 Gene (382,6 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, ALDOA, AMPD1, ANO5, BAG3, BIN1, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, ECEL1, ENO3, ETFA, ETFB, ETFDH, FLAD1, FLNC, FXR1, GAA, GBE1, GNE, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LDB3, LAMP2, LDHA, LIG3, LMOD3, LPIN1, MAP3K20, MATR3, MEGF10, MGME1, MTM1, MTMR14, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYMK, MYOT, MYPN, NEB, TNNT1, OPA1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, PYROXD1, RBCK1, RRM2B, RYR1, SELENON, SLC22A5, SLC25A20, SLC25A4, SPEG, STAC3, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TIA1, TK2, TNNT1, TNNT3, TPM2, TPM3, TTN, TYMP, UBA1, YARS2 Gliedergürtelmuskeldystrophie (LGMD) und weitere Muskeldystrophien: 47 Gene (270,0 kb) ANO5, B3GALNT2, B4GAT1, BVES, CAPN3, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, HNRNPDL, LAMA2, LARGE1, LIMS2, LMNA, PLEC, POGlut1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RXYLT1, SGCA, SGCB, SGC, SGCG, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
Muskelerkrankungen mit Herzbeteiligung * Gen-Panel: ID123.03 Muskelerkrankungen mit Herzbeteiligung: 19 Gene (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss-Muskeldystrophie (EMDM): 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker-Muskeldystrophie (DMD, BMD): 1 Gen (11,1 kb) DMD	4 - 6 Wo	E
Myofibrilläre Myopathie (MFM) * Gen-Panel: ID161.01, 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B	4 - 6 Wo	E
Myotonie * Gen-Panel: ID255.00, 5 Gene (25,0 kb) ATP2A1, CLCN1, HINT1, HSPG2, SCN4A	3 - 5 Wo	E
Nemalin-Myopathie (NEM) * Gen-Panel: ID199.00, 11 Gene (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3	3 - 5 Wo	E
Periodische Paralyse * Gen-Panel: ID253.00, 7 Gene (16,6 kb) CACNA1S, KCNE3, KCNJ2, KCNJ5, KCNJ12, KCNJ18, SCN4A	3 - 5 Wo	E
Spinale Muskelatrophie (SMA) * Gen-Panel: ID152.02 Spinale Muskelatrophie (SMA): 39 Gene (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1 Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB Distale spinale Muskelatrophie (DSMA, HMN): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	3 - 5 Wo	E
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPBP, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Zentronukleäre Myopathie (CNM) * Gen-Panel: ID257.00, 7 Gene (21,4 kb) CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG	3 - 5 Wo	E
Neurodegenerative Erkrankungen		
Alzheimer-Krankheit (AD) * Gen-Panel: ID157.01, 7 Gene (16,8 kb) ABCA7, ADAM10, APOE, APP, PSEN1, PSEN2, TF	3 - 5 Wo	E
Amyotrophe Lateralsklerose (ALS) * Gen-Panel: ID209.03, 35 Gene (80,2 kb) ALS2, ANG, ANXA11, ATXN2, C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, LRP12, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SPTLC1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP	3 - 5 Wo	E
Basalganglien-Kalzifikation (IBGC) * Gen-Panel: ID327.00, 6 Gene (11,2 kb) JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1	3 - 5 Wo	E
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN) * Gen-Panel: ID052.03 Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN): 45 Gene (111,8 kb) AARS1, AIFM1, ATP1A1, CADM3, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, JAG1, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PNKP, PRPS1, RAB7A, SLC12A6, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth-Neuropathie, axonal, dominant (CMT2): 26 Gene (74,2 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, JAG1, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, SLC12A6, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, axonal, rezessiv (CMT2): 10 Gene (24,4 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MFN2, MME, MPV17, PNKP, SPG11, TRIM2 Charcot-Marie-Tooth-Neuropathie, Intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Charcot-Marie-Tooth-Neuropathie mit Optikusatrophy (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN) * 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	3 - 5 Wo	E
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik * Gen-Panel: ID312.02 Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik: 87 Gene (217,3 kb) AARS1, AIFM1, ARHGEF10, ATP1A1, ATP7A, BSCL2, CADM3, CNTNAP1, COQ7, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SORD, SPG11, SPTAN1, SPTLC1, SURF1, SYT2, TRIM2, TRPV4, VCP, VRK1, VWA1, WARS1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2): 35 Gene (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophe Déjerine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Charcot-Marie-Tooth-Neuropathie mit Optikusatrophy (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46 Distale motorische Neuropathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 CADASIL und CARASIL *, # Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	4 - 6 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, RNFB216, SLC2A1, VPS13A, XK	3 - 5 Wo	E
Distale hereditäre motorische Neuronopathie (HMND, HMNR) * Gen-Panel: ID254.01 Distale hereditäre motorische Neuronopathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Distale motorische Neuronopathie, autosomal-dominant (HMND): 15 Gene (38,4 kb) BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1 Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 10 Gene (14,4 kb) ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1	3 - 5 Wo	E
Dystonie (DYT) * Gen-Panel: ID128.04 Dystonie (DYT): 25 Gene (55,9 kb) ACTB, ADCY5, ANO3, AOEPE, 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, AOEPE, 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

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Episodisches Schmerzsyndrom (FEPS) * Gen-Panel: ID268.00, 4 Gene (20,5 kb) SCN9A, SCN10A, SCN11A, TRPA1	3 - 5 Wo	E
Essentieller Tremor (ETM) * Gen-Panel: ID195.01, 4 Gene (16,6 kb) DRD3, FUS, SCN4A, TENM4	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
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
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, umfassende Diagnostik * Gen-Panel: ID374.00 Hereditäre Neuropathien, umfassende Diagnostik: 134 Gene (343,6 kb) AARS1, ABCA1, ACOX1, AIFM1, AR, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BICD2, BSCL2, CADM3, CFAP276, CHCHD10, CNTNAP1, COQ7, COX20, COX6A1, CPOX, CYP27A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, GSN, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MAG, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PDXK, PIGB, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PPOX, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN9A, SCN10A, SCN11A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A6, SLC5A7, SLC52A2, SLC52A3, SMN1, SORD, SPG11, SPTAN1, SPTLC1, SPTLC2, SURF1, SYT2, TECPR2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VCP, VRK1, VWA1, WARS1, WNK1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2, HMSN2): 35 Gene (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX, HMSN): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophe Déjerine-Sottas-Neuropathie (DSS, HMSN3): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb) EGR2, MPZ, CNTNAP1 Distale motorische Neuropathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Sensorische und autonome Neuropathie (HSAN, HSN): 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Hereditäre sensorische und autonome Neuropathie (HSAN, HSN) * Gen-Panel: ID086.02, 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	3 - 5 Wo	E
Hirnatrophie und demyelinisierende Erkrankungen des Gehirns * Gen-Panel: ID278.00 Hirnatrophie und demyelinisierende Erkrankungen des Gehirns: 55 Gene (96,0 kb) AIMP1, AIMP2, ASPA, B3GALNT2, B4GAT1, CNP, COL4A1, CRPPA, DAG1, DARS1, DEGS1, EPRS1, FAM126A, FARSA, FARSB, FKRP, FKTN, GFAP, GJC2, GMPPB, GRM7, HIKESHI, HSPD1, LARGE1, MAPT, MAT1A, MED17, MTHFS, PLP1, POLR1C, POLR3A, , POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSEN1, PYCR2, RARS1, RXYLT1, SLC25A12, SOX10, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Hirnatrophie: 13 Gene (24,2 kb) EXOC7, EXOC8, FARSA, FARSB, GRM7, MAPT, MED17, PSEN1, TBCD, TRAPPC4, TRAPPC6B, TRAPPC12, UBTf Hypo- und Demyelinisierung des Gehirns: 27 Gene (43,0 kb) AIMP1, AIMP2, ASPA, CNP, DARS1, DEGS1, EPRS1, FAM126A, GFAP, GJC2, HIKESHI, HSPD1, MAT1A, MTHFS, PLP1, POLR1C, POLR3A, POLR3B, PYCR2, RARS1, SLC25A12, SOX10, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Walker-Warburg-Syndrom (MDDGA): 14 Gene (23,6 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Hyperekplexie (HKPX) * Gen-Panel: ID216.00, 9 Gene (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 Wo	E
Hypomyelinisierende Leukodystrophie (HLD) * Gen-Panel: ID277.00, 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11	3 - 5 Wo	E
Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik * Gen-Panel: ID204.04 Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik: 122 Gene (208,5 kb) AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, AIFM1, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, AUH, BOLA3, CLCN2, CLDN11, CNP, COA7, COA8, COL4A1, COL4A2, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS1, DARS2, DEGS1, EARS2, EIF2AK1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FA2H, FAM126A, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GCDH, GFAP, GFM1, GJC2, GLB1, HEPACAM, HIKESHI, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, ISCA1, ISCA2, KARS1, KCNT1, L2HGDH, LMNB1, LYRM7, MARS2, MCOLN1, MLC1, MTFMT, NAXD, NAXE, NFU1, NKX6-2, NOTCH3, NUBPL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PLAA, PLEKHG2, PLP1, PMPCB, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SDHAF1, SLC13A3, SLC16A2, SLC17A5, SOX10, SPTAN1, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYMP, TYROBP, UFM1, VPS11, ZFYVE26 Leukodystrophie mit Hypomyelinisierung (HLD): 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Leukodystrophie mit Peroxisomenbiogenese-Störung (PBD): 15 Gene (20,8 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH Orthochromatische Leukodystrophie: 10 Gene (14,6 kb) ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1 Metachromatische Leukodystrophie: 3 Gene (4,2 kb) ARSA, PSAP, SUMF1 Aicardi-Goutières-Syndrom (AGS): 7 Gene (11,9 kb) ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1 CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3	4 - 6 Wo	E
Neurodegeneration mit Eisenablagerung im Gehirn (NBIA) * Gen-Panel: ID264.00, 11 Gene (20,0 kb) ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45	3 - 5 Wo	E
Neurologische Entwicklungsstörungen (NED), umfassende Diagnostik * Gen-Panel: ID358.00, 169 Gene (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6VOA1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNA1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDF1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNP1, HNRNP, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNP, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAP1, RBL2, RERE, SRS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNF, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 Wo	E
Neuronale Ceroid-Lipofuszinose (CLN) * Gen-Panel: ID132.01, 15 Gene (20,2 kb) ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1	3 - 5 Wo	E
Paroxysmale Dyskinesie * Gen-Panel: ID286.00, 14 Gene (38,9 kb) ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Parkinson-Krankheit (PARK) * Gen-Panel: ID077.02 Parkinson-Krankheit (PARK): 37 Gene (93,8 kb) ADH1C, ATP13A2, ATP1A3, ATP6AP2, CHCHD2, DCTN1, DNAJC6, EIF4G1, FBXO7, GBA, GCH1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, POLG, PRKRA, PRKN, PSAP, RAB39B, SLC18A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SYNJ1, TAF1, TH, TRPM7, UCHL1, UQCRC1, VPS13C, VPS35 Parkinson-Krankheit (PARK), early-onset: 9 Gene (30,3 kb) ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, SYNJ1, VPS13C Parkinson-Krankheit (PARK), late-onset: 12 Gene (27,3 kb) ADH1C, CHCHD2, EIF4G1, GBA, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, SNCA, UCHL1, VPS35 Dystonie-Parkinsonismus (DYT): 9 Gene (18,3 kb) ATP1A3, GCH1, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH	3 - 5 Wo	E
Schlafstörungen * Gen-Panel: ID371.00, 13 Gene (25,3 kb) ADRB1, BHLHE41, CRY1, CRY2, CSNK1D, GRM1, HCRT, MOG, NPSR1, PER2, PER3, PRNP, TIMELESS	3 - 5 Wo	E
Spastische Ataxie (SPAX) * Gen-Panel: ID228.00, 12 Gene (34,8 kb) AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1	3 - 5 Wo	E
Spastische Paraplegie (HSP, SPG) * Gen-Panel: ID148.04 Spastische Paraplegie (HSP, SPG): 67 Gene (138,1 kb) ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27 Spastische Paraplegie (SPG), autosomal-dominant: 20 Gene (43,1 kb) ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27 Spastische Paraplegie (SPG), autosomal-rezessiv: 50 Gene (101,2 kb) ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, UCHL1, VPS37A, ZFYVE26 Spastische Paraplegie (SPG), X-chromosomal: 3 Gene (6,9 kb) ABCD1, L1CAM, PLP1	4 - 6 Wo	E
Spinale Muskelatrophie (SMA) * Gen-Panel: ID152.02 Spinale Muskelatrophie (SMA): 39 Gene (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1 Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB Distale spinale Muskelatrophie (DSMA, HMN): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	3 - 5 Wo	E
Spinozerebelläre Ataxie, autosomal-dominant (SCA, ADCA) * 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	3 - 5 Wo	E
Spinozerebelläre Ataxie, autosomal-rezessiv (SCAR, SCAN) * 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	4 - 6 Wo	E
Störung der Peroxisomenbiogenese (PBD) * Gen-Panel: ID083.01 Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 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	3 - 5 Wo	E

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

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Hyperoxalurie Gen-Panel: ID363.00 Hyperoxalurie: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb) AGXT, GRHPR, HOGA1 Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 Wo	E
Hypophosphatasie, Hypophosphatämie und Rachitis * Gen-Panel: ID269.03 Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb) ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb) CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3 Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb) CYP2R1, CYP3A4, CYP27B1, VDR Hypophosphatasie (HPP): 1 Gen (1,6 kb) ALPL	3 - 5 Wo	E
Joubert-Syndrom (JBTS) * Gen-Panel: ID028.03, 40 Gene (104,1 kb) AH11, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 Wo	E
Kongenitale Anomalien der Niere und ableitenden Harnwege (CAKUT) * Gen-Panel: ID229.03 Kongenitale Anomalien der Niere und ableitenden Harnwege (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 Branchiotoxikales Syndrom (BOR): 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A Renale tubuläre Dysgenese (RTD): 4 Gene (7,7 kb) ACE, AGT, AGTR1, REN Fraser-Syndrom (FRAS): 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1 MMIH-Syndrom (MMIH): 5 Gene (15,1 kb) ACTG2, LMOD1, MYH11, MYL9, MYLK	4 - 6 Wo	E
Meckel-Syndrom (MKS) * Gen-Panel: ID032.02, 13 Gene (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 Wo	E
Metabolische Nierenerkrankungen * Gen-Panel: ID705.00, 29 Gene (53,4 kb) AGXT, APOA1, ATP7B, B2M, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, FAH, FGA, GALT, GLA, GRHPR, GSN, HOGA1, KCNJ1, LYZ, MEFV, MVK, NLRC4, NLRP12, NLRP3, OCRL, PLCG2, SLC12A1, SLC26A1, TNFRSF1A, TTR	3 - 5 Wo	E
Mikrohämaturie * Gen-Panel: ID385.00, 11 Gene (44,0 kb) CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FN1, INF2, MYH9, PIGA, PIGT, UMOD	3 - 5 Wo	E
Nephroblastom und Wilms-Tumor (WT) * Gen-Panel: ID335.01, 30 Gene (97,7 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CHEK2, CTR9, DICER1, DIS3L2, FBXW7, FIBP, GPC3, GPC4, KDM3B, NSD1, NYN1RIN, MLH1, MSH2, MSH6, PALB2, PMS2, POU6F2, REST, TP53, TRIM28, TRIM37, TRIP13, WT1	3 - 5 Wo	E
Nephrokalzinose * Gen-Panel: ID361.00, 29 Gene (54,1 kb) ADCY10, AGXT, ALPL, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, FAH, FAM20A, GRHPR, HOGA1, KCNJ1, MAGED2, OCRL, OXGR1, RRAGD, SLC12A1, SLC4A1, SLC34A1, SLC34A3, VIPAS39, VPS33B	3 - 5 Wo	E
Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS) * Gen-Panel: ID098.06 Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS): 43 Gene (129,7 kb) ACTN4, ANLN, APOL1, ARHGAP24, ARHGAP24, 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, ARHGAP24, AVIL, COQ2, COQ6, COQ8B, DAAM2, DGKE, EMP2, KANK2, KIRREL1, LAMA5, LAMB2, MAGI2, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, WT1 Fokal-segmentale Glomerulosklerose (FSGS): 18 Gene (49,5 kb) ACTN4, ANLN, APOL1, ARHGAP24, COQ2, COQ6, COL4A3, COL4A4, COL4A5, CRB2, CD2AP, INF2, LMX1B, MYO1E, PAX2, PDSS2, SMARCAL1, TRPC6	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Nephronophthise (NPHP) * Gen-Panel: ID030.02, 22 Gene (70,9 kb) ANKS6, CEP83, CEP164, CEP290, DDC2C, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	3 - 5 Wo	E
Nierenzellkarzinom * Gen-Panel: ID041.04, 34 Gene (76,8 kb) BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1	3 - 5 Wo	E
Polyzystische Lebererkrankung (PCLD) * Gen-Panel: ID305.02 Polyzystische Lebererkrankung (PCLD): 16 Gene (55,7 kb) ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63 Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 7 Gene (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 9 Gene (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1	3 - 5 Wo	E
Polyzystische Nierenerkrankung (PKD) * Gen-Panel: ID295.03 Polyzystische Nierenerkrankung (PKD): 9 Gene (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1 Polyzystische Nierenerkrankung, autosomal-dominant (ADPKD): 7 Gene (27,4 kb) ALG5, DNAJB11, GANAB, IFT140, NEK8, PKD1, PKD2 Polyzystische Nierenerkrankung, autosomal-rezessiv (ARPKD): 2 Gene (14,5 kb) DZIP1L, PKHD1	3 - 5 Wo	E
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) * Gen-Panel: ID250.01 Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA): 8 Gene (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 Wo	E
Renale Zilopathien, umfassende Diagnostik * Gen-Panel: ID376.00 Renale Zilopathien, umfassende Diagnostik: 75 Gene (181,7 kb) AH11, 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, DDC2C, 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, DDC2C, 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) AH11, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423 Meckel-Syndrom (MKS): 14 Gene (36,2 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15	4 - 6 Wo	E
Renale Amyloidose * Gen-Panel ID320.01 Renale Amyloidose: 9 Gene (13,2 kb) APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 6 Gene (6,3 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Periodisches Fiebersyndrom mit Amyloidose: 3 Gene (6,8 kb) MEFV, NLRP3, TNFRSF1A	3 - 5 Wo	E
Renale Hypodysplasie, Aplasie und Agenesie * Gen-Panel: ID319.00, 23 Gene (72,5 kb) BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4	3 - 5 Wo	E
Renale tubuläre Azidose (RTA) * Gen-Panel: ID297.00, 9 Gene (18,5 kb) ATP6VOA4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Renale tubuläre Dysgenese (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 Thrombotische Mikroangiopathie (TMA): 23 Gene (44,7 kb) ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD Thrombotisch-thrombozytopenische Purpura (TTP): 1 Gen (4,3 kb) ADAMTS13 Atypisches hämolytisch-urämisches Syndrom (AHUS): 23 Gene (44,7 kb) ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD Tubulointerstitielle Nierenerkrankung, autosomal-dominant (ADTKD) * Gen-Panel: ID296.00, 6 Gene (8,8 kb) DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD	3 - 5 Wo	E
Urolithiasis, Nephrolithiasis und Nephrokalzinose * Gen-Panel ID231.04 Urolithiasis, Nephrolithiasis und Nephrokalzinose: 37 Gene (68,8 kb) ADCY10, AGXT, ALPL, APRT, ATP6VOA4, ATP6V1B1, BSND, CASR, CLCN5, CLDN16, CLDN19, CLCNKB, CYP24A1, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, OCRL, OXGR1, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, XDH Hyperkalziurie: 15 Gene (30,2 kb) ADCY10, BSND, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, RRAGD, SLC12A1, SLC34A1, SLC34A3 Hyperoxalurie: 5 Gene (6,3 kb) AGXT, GRHPR, HOGA1, SLC26A1, OXGR1 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Hypocitraturie: 4 Gene (11,1 kb) TP6VOA4, ATP6V1B1, SLC4A1, SLC4A4 Xanthinurie: 2 Gene (6,7 kb) MOCOS, XDH Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9	3 - 5 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
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

Erkrankung/Diagnostik	Dauer	Material
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, TGFB1, TGFB2, 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, RAPSIN, 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, RAPSIN	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, EFN1, FGFR1, FGFR2, GLI3, TWIST1, ZSWIM6	3 - 5 Wo	E
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, EFN1, 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, WPCP, 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

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Herz-Hand-Syndrom * Gen-Panel: ID165.01, 9 Gene (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 Wo	E
Hypophosphatasie, Hypophosphatämie und Rachitis * Gen-Panel: ID269.03 Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb) ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb) CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3 Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb) CYP2R1, CYP3A4, CYP27B1, VDR Hypophosphatasie (HPP): 1 Gen (1,6 kb) ALPL	3 - 5 Wo	E
Klippel-Feil-Syndrom (KFS) * Gen-Panel: ID207.00, 5 Gene (12,5 kb) GDF3, GDF6, MEOX1, MYO18B, PAX1	3 - 5 Wo	E
Kraniosynostose (CRS) * Gen-Panel: ID224.02 Kraniosynostose (CRS): 36 Gene (84,9 kb) ALPL, ALX4, ASXL1, CD96, CDC45, COLEC10, COLEC11, CYP26B1, EFN1, 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 Trigonocephalie (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, DYNC2L1, 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, DYNC2L1, 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 Kontrakturnsyndrom (LCCS) * Gen-Panel: ID197.00, 12 Gene (34,6 kb) ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42	3 - 5 Wo	E
Lippen-, Kiefer- und Gaumenspalte (OFC, CLP) * Gen-Panel: ID266.01 Lippen-, Kiefer- und Gaumenspalte (OFC, CLP): 50 Gene (151,0 kb) ARHGAP29, BMP4, BRD4, C2CD3, CDH1, CHD7, CDH11, CPLANE1, CYFIP1, DDX59, DHCR7, DLX4, ESCO2, FGFR1, FGFR2, FLNB, GRHL3, HDAC8, IFT57, INTU, IRF6, KDM1A, KDM6A, KIAA0753, KMT2D, LRRC32, MEIS2, MID1, MSX1, NECTIN1, NEK1, NIPA1, NIPA2, NIPBL, OFD1, RAD21, RAB34, RIPK4, SCN11, 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, SCN11, TCTN3, TMEM107, ZRSR2 Cornelia-de-Lange-Syndrom (CDLS): 6 Gene (22,9 kb) BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3 Kabuki-Syndrom (KABUK): 3 Gene (23,5 kb) KDM1A, KDM6A, KMT2D Van-der-Woude-Syndrom (VWS): 2 Gene (3,3 kb) GRHL3, IRF6	4 - 6 Wo	E
Mandibulofaziale Dysostose (MFD) * Gen-Panel: ID188.01 Mandibulofaziale Dysostose (MFD): 11 Gene (22,0 kb) DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D, RPS28, SF3B4, TCOF1, TSR2 Treacher-Collins-Syndrom (TCS): 4 Gene (9,4 kb) POLR1B, POLR1C, POLR1D, TCOF1	3 - 5 Wo	E
Multiple epiphysäre Dysplasie (EDM) * Gen-Panel: ID202.02, 11 Gene (27,7 kb) CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CSGALNACT1, EIF2AK3, KIF7, MATN3, SLC26A2	3 - 5 Wo	E
Multiples Pterygium-Syndrom * Gen-Panel: ID158.01, 8 Gene (16,8 kb) CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Orofaziodigitales Syndrom (OFD) * Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E
Osteogenesis imperfecta (OI) * Gen-Panel: ID066.02, 21 Gene (39,2 kb) ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1	3 - 5 Wo	E
Osteopetrose (OPT) und ähnliche Knochenerkrankungen * Gen-Panel ID346.01 Osteopetrose (OPT) und ähnliche Knochenerkrankungen: 32 Gene (68,1 kb) AMER1, ANKH, CA2, CLCN7, CSF1R, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, LRP6, LRRK1, OSTM1, PLEKHM1, PTDSS1, PTH1R, SLC4A2, SLC29A3, SLC02A1, 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, SLC02A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP	3 - 5 Wo	E
Osteoporose * Gen-Panel: ID115.02, 15 Gene (39,0 kb) CALCR, COPB2, COL1A1, COL1A2, ESR1, LGR4, LRP5, NOTCH2, PLS3, NHERF1, SGMS2, SLC34A1, VDR, WNT1, WNT11	3 - 5 Wo	E
Pierre-Robin-Sequenz * Gen-Panel: ID294.00, 33 Gene (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRNP, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E
Polydaktylie, nicht-syndromale Form * Gen-Panel: ID166.02, 9 Gene (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141	3 - 5 Wo	E
Rubinstein-Taybi-Syndrom (RSTS) * Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
Skelettdysplasie, schwere Form * Gen-Panel: ID056.01 Skelettdysplasie, schwere Form: 46 Gene (144,2 kb) AGPS, ALPL, BMPER, CANT1, CEP120, CILK1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DLL3, DYNC2H1, EBP, FAM20C, FAM111A, FGFR2, FGFR3, FLNA, FLNB, GDF5, GNPAT, GPX4, HSPG2, IFT52, IFT80, IFT81, INPPL1, KIAA0586, LBR, LIFR, NEK1, NSDHL, P3H1, PEX5, PEX7, PPIB, PTH1R, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4, WDR34, WDR35 Achondrogenesie (ACG): 4 Gene (14,1 kb) COL2A1, GDF5, SLC26A2, TRIP11 Fibrochondrogenesie (FBCG): 2 Gene (10,6 kb) COL11A1, COL11A2 Thanatophore Dysplasie (TD): 2 Gene (6,9 kb) COL1A2, FGFR3 Chondrodysplasie, letal: 12 Gene (24,5 kb) AGPS, EBP, FLNB, GDF5, GNPAT, GPX4, LBR, PEX5, PEX7, PTH1R, SLC26A2, SLC35D1 Osteogenesis imperfecta (OI), letal: 5 Gene (12,6 kb) CRTAP, COL1A1, COL1A2, P3H1, PPIB Kurzrippen-Thoraxdysplasie (SRTD), letal: 9 Gene (35,0 kb) CEP120, DYNC2H1, IFT52, IFT80, IFT81, KIAA0586, NEK1, WDR34, WDR35	4 - 6 Wo	E
Skelettdysplasien, umfassende Diagnostik * Gen-Panel: ID356.00, 407 Gene (969,6 kb) ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF3, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARL6, ARSB, ARSL, ASXL1, ASXL2, ATP6VOA2, ATP7A, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CDC8, 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, FGF19, FGF2, FGF3, FGF4, 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, RPRIP1L, 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, SLC02A1, SMAD3, SMAD4, SMAD6, SMARCA1, SMC1A, SMC3, SMOC1, SNRNP, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFB3, TMC01, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UFSP2, UNC45A, VDR, WBP11, WPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenenerkrankungen		
Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD) * Gen-Panel: ID110.01 Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD): 39 Gene (90,7 kb) ACAN, ACP5, AIFM1, B3GALT6, BGN, CFAP410, CHST3, COL2A1, COL10A1, COL11A2, COMP, DDR2, DDRGK1, EXOC6B, FN1, GPX4, KIF22, MATN3, MBTPS1, MMP13, NANS, NEPRO, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, PLCB3, POP1, RPL13, RSPRY1, SIK3, SMARCAL1, TONSL, TRAPPC2, TRIP11, TRPV4, UFSP2, WISP3 Spondyloepiphysäre Dysplasie (SED): 9 Gene (25,9 kb) ACAN, CHST3, COL2A1, COMP, MBTPS1, SMARCAL1, TRAPPC2, TRPV4, WISP3 Spondylometaphysäre Dysplasie (SMD): 11 Gene (30,2 kb) ACP5, CFAP410, COL2A1, COL10A1, FN1, GPX4, PAM16, PCYT1A, PLCB3, TRIP11, TRPV4 Spondyloepimetaphysäre Dysplasie (SEMD): 21 Gene (47,6 kb) ACAN, AIFM1, B3GALT6, BGN, COL2A1, DDR2, DDRGK1, EXOC6B, KIF22, MATN3, MMP13, NANS, NEPRO, PAPSS2, PISD, POP1, RPL13, RSPRY1, SIK3, TONSL, UFSP2	3 - 5 Wo	E
Spondylkostale Dysostose (SCDO) * Gen-Panel: ID227.00, 7 Gene (14,4 kb) DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6	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
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
Coenzym-Q10-Mangel (COQ10D) * Gen-Panel: ID225.01, 15 Gene (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2	3 - 5 Wo	E
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen * Gen-Panel: ID706.01, 16 Gene (30,1 kb) ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1	3 - 5 Wo	E
Folatstoffwechselstörung * Gen-Panel: ID334.00, 10 Gene (18,7 kb) CBS*, FOLR1, FOLR2, FTHD, MTHFD1, MTHFR, MTR, MTRR, SLC19A1, SLC46A1	3 - 5 Wo	E
Glykogenspeicherkrankheit (GSD) * Gen-Panel: ID108.01, 29 Gene (54,1 kb) AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC1, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4	3 - 5 Wo	E
Glykosylphosphatidylinositol (GPI)-Biosynthesedefekt (GPIBD) * Gen-Panel: ID291.00 Glykosylphosphatidylinositol(GPI)-Biosynthesedefekt (GPIBD): 22 Gene (33,3 kb) GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY Multiple kongenitale Anomalien-Hypotonie-Krampfanfälle-Syndrom (MCAHS): 4 Gene (7,7 kb) PIGA, PIGN, PIGQ, PIGT Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS): 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
Hämochromatose (HFE) und Häm siderose * Gen-Panel: ID114.04, 10 Gene (14,6 kb) BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Homocystinurie * Gen-Panel: ID191.01, 9 Gene (15,3 kb) ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1	3 - 5 Wo	E
Hyperinsulinämische Hypoglykämie (HHF) * Gen-Panel: ID126.00, 8 Gene (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 Wo	E
Hyperkalzämie * Gen-Panel: ID262.00, 8 Gene (14,6 kb) AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1	3 - 5 Wo	E
Hyperoxalurie Gen-Panel: ID363.00 Hyperoxalurie: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb) AGXT, GRHPR, HOGA1 Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 Wo	E
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) * Gen-Panel: ID292.00, 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung * Gen-Panel: ID280.00 Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung: 44 Gene (85,9 kb) ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4 Glykogenspeicherkrankheit (GSD): 24 Gene (48,4 kb) AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4 Hyperinsulinämische Hypoglykämie (HHF): 9 Gene (18,8 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A	3 - 5 Wo	E
Hypomagnesiämie (HOMG) * Gen-Panel ID054.02: 14 Gene (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYP2, HNF1B, KCNA1, KCNJ10, RRGD, SLC12A3, TRPM6	3 - 5 Wo	E
Hypophosphatasie, Hypophosphatämie und Rachitis * Gen-Panel: ID269.03 Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb) ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb) CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3 Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb) CYP2R1, CYP3A4, CYP27B1, VDR Hypophosphatasie (HPP): 1 Gen (1,6 kb) ALPL	3 - 5 Wo	E
Kombinierter Defekt der oxidativen Phosphorylierung (COXPD) * Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPPE, 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, PTCO3, 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, ATP6VOA2, 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, ATP6VOA2, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAN2B2, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Kongenitale Glykosylierungsstörung, Typ II (CDG2): 27 Gene (42,7 kb) ATP6AP1, ATP6AP2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, GET4, MAN1B1, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, STX5, TMEM165, VMA12, VMA22	3 - 5 Wo	E
MODY-Diabetes * Gen-Panel: ID048.01, 14 Gene (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 Wo	E
Mukopolysaccharidose (MPS) * Gen-Panel: ID308.00, 12 Gene (21,2 kb) ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Muskeldystrophie-Dystroglykanopathie (MDDG) * 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
Metabolische Epilepsien * Gen-Panel: ID303.01 Metabolische Epilepsien: 84 Gene (122,3 kb) ABAT, ACY1, ADL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNM2, 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
Neonataler Diabetes mellitus * Panel: ID162.01 Neonataler Diabetes mellitus: 29 Gene (53,6 kb) ABCC8, BSLC2, 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) BSLC2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 Wo	E
Neuronale Ceroid-Lipofuszinose (CLN) * Gen-Panel: ID132.01, 15 Gene (20,2 kb) ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1	3 - 5 Wo	E
Porphyrrie * Gen-Panel: ID153.01, 10 Gene (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 Wo	E
Pulmonale Surfactant-Stoffwechselstörung (SMDP) * Gen-Panel: ID168.01, 6 Gene (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 Wo	E
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
Speicherkrankheiten mit Herzbeteiligung * 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	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Stoffwechselstörung mit Epilepsie im Neugeborenenalter * Gen-Panel: ID135.00, 25 Gene (39,4 kb) ABAT, ADL, 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
Störung der Peroxisomenbiogenese (PBD) * Gen-Panel: ID083.01 Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 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	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
3-Methylglutaconazidurie (MGCA) * 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	3 - 5 Wo	E
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, RFW3, 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, HLT, 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, PARBP, 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, RFW3, 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
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, RFW3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Erbliche Tumorerkrankungen, umfassende Diagnostik * Gen-Panel: ID018.04, 353 Gene (779,1 kb) ABRAXAS1, ACD, ADA2, ADH5, AIP, AKT1, ALDH2, ALK, AMER1, ANAPC1, ANKRD26, APC, APTX, AR, ARID1A, ASXL1, ATM, ATR, ATRX, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BTK, BUB1B, CASP10, CASP8, CASR, CBL, CCND1, CDC73, CDCA7, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CEBPA, CEP57, CHEK2, CLPB, CSF3R, CTC1, CTLA4, CTNNA1, CTR9, CTRC, CYLD, CYP3A43, DCLRE1B, DCLRE1C, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNA2, DNAJC21, DNMT3A, DNMT3B, DUT, EFL1, EGFR, EHBP1, ELAC2, ELANE, ELP1, ENOSF1, EPCAM, ERBB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ETV6, EXO1, EXT1, EXT2, EZH2, FAH, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGFR3, FH, FLCN, FOXE1, G6PC3, GALNT12, GATA1, GATA2, GDNF, GEN1, GF11, GPC3, GPC4, GREM1, GTF2E2, GTF2H5, HABP2, HAVCR2, HAX1, HCLSL1, 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, TGFB2, TINF2, TMEM127, TNFRSF11A, TOP3A, TP53, TREX1, TRIM28, TRIM37, TRIP13, TRRAP, TSC1, TSC2, TSR2, TYMS, TYR, UBE2A, UBE2T, UNC13D, UNG, USB1, USP45, UVSSA, VHL, VPS45, WAS, WRAP53, WRN, WT1, WWP1, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, ZBTB24, ZCCHC8, ZNF687, ZSWIM7	4 - 6 Wo	E
Gastrointestinaler Stromatumor (GIST) * Gen-Panel: ID226.00, 8 Gene (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 Wo	E
Glioblastom (GLM) * Gen-Panel: ID313.00, 16 Gene (61,0 kb) APC, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POT1, PTEN, TP53, TSC1, TSC2	3 - 5 Wo	E
Gorlin-Syndrom (BCNS) und ähnliche Krankheitsbilder * Gen-Panel: ID174.02, 8 Gene (25,8 kb) BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 Wo	E
Hypophysenadenom (PITA) * Gen-Panel: ID387.01, 21 Gene (47,3 kb) AIP, CDH23, CDKN1B, DICER1, EPCAM, GNAS, GPR101, MAX, MLH1, MSH2, MSH6, MEN1, PMS2, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, USP8	3 - 5 Wo	E
Kolorektales Karzinom und Polyposis* Gen-Panel ID006.09 Kolorektales Karzinom und Polyposis: 23 Gene (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Kolorektales Karzinom: 18 Gene (55,7 kb) APC, ATM, CHEK2, BMPR1A, EPCAM, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11 Kolorektale Polyposis: 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
Kolorektales Karzinom mit Mikrosatelliteninstabilität (MSI) * Gen-Panel: ID283.00, 9 Gene (25,4 kb) EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE	3 - 5 Wo	E
Krebserkrankungen im Kindesalter * Gen-Panel: ID333.00 Krebserkrankungen im Kindesalter: 139 Gene (341,1kb) ACD, ALK, ANKRD26, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNAJC21, EFL1, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, GATA2, GPC3, HRAS, IKZF1, KIF1B, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PMS2, POLE, POLH, POU6F2, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RAD51, RAD51C, RAF1, RB1, RECQL4, REST, RET, RFWD3, RIT1, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, STK11, SUFU, TERT, TMEM127, TINF2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2 Maligne hämatologische Erkrankungen: 49 Gene (126,4 kb) ACD, ANKRD26, ATM, BLM, BRCA1, BRCA2, BRIP1, CEBPA, DDX41, DKC1, DNAJC21, EFL1, ELANE, ERCC4, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA2, IKZF1, MAD2L2, NBN, NHP2, NOP10, PALB2, PARN, PAX5, RAD51, RAD51C, RFWD3, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SRP72, TERT, TINF2, TP53, UBE2T, WRAP53, XRCC2 Tumoren des Zentralnervensystems: 32 Gene (111,9 kb) ALK, APC, BRCA2, CDKN2A, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, KIF1B, LZTR1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PALB2, PHOX2B, PMS2, PTCH1, PTCH2, PTEN, RB1, SMARCA4, SMARCB1, SUFU, TP53, TSC1, TSC2, VHL Endokrine Tumoren: 15 Gene (21,2 kb) CDC73, CDKN1B, DLST, KIF1B, MAX, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL RASopathien: 18 Gene (36,6 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2 Wilms-Tumor (WT): 10 Gene (29,5 kb) BRCA2, CDKN1C, GPC3, DIS3L2, POU6F2, TRIM28, WT1, TRIM37, CDC73, REST Xeroderma pigmentosum (XP): 9 Gene (19,0 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC MMR-Defizienz-Syndrom (MMRCS): 5 Gene (12,7 kb) MLH1, MSH2, PMS2, MSH6, EPCAM	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Konstitutionelles MMR-Defizienz-Syndrom (CMMRDS, MMRCS) * Gen-Panel: ID362.00, 5 Gene (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	2 - 4 Wo	E
Kutanes malignes Melanom (CMM) * Gen-Panel: ID193.01, 12 Gene (26,4 kb) BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3	3 - 5 Wo	E
Lynch-Syndrom (LYNCH, HNPCC) * Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	3 - 5 Wo	E
Lungenkarzinom * Gen-Panel: ID260.02, 33 Gene (122,5 kb) ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTHY, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53	4 - 6 Wo	E
Magenkarzinom * Gen-Panel: ID090.04, 25 Gene (73,6 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTHY, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	3 - 5 Wo	E
Mammakarzinom * Gen-Panel: ID021.02, 12 Gene (41,0 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53	3 - 5 Wo	E
Mamma- und Ovarialkarzinom (HBOC) * Gen-Panel: ID003.04 Mamma- und Ovarialkarzinom (HBOC): 19 Gene (62,5 kb) ATM, BARD1, BRIP1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53 Mammakarzinom: 10 Gene (38,9 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, STK11, TP53 Ovarialkarzinom: 14 Gene (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 Wo	E
Medulloblastom (MDB) * Gen-Panel: ID205.02, 22 Gene (75,7 kb) APC, BRCA2, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTCH1, PTCH2, PTEN, SMARCB1, SMARCA4, SUFU, TP53, VHL	3 - 5 Wo	E
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) * Gen-Panel: ID321.01 Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML): 121 Gene (244,1 kb): ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8 Akute myeloische Leukämie (AML): 12 Gene (28,4 kb) ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53 Diamond-Blackfan-Anämie (DBA): 20 Gene (11,3 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Shwachman-Diamond-Syndrom (SDS): 4 Gene (7,2 kb) DNAJC21, EFL1, SBDS, SRP54 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Lungenfibrose und Knochenmarkinsuffizienz (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8 Dyskeratosis congenita (DKC): 13 Gene (21,8 kb) ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53 Kongenitale Neutropenie (SCN): 10 Gene (13,9 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS Fanconi-Anämie (FANC): 20 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2 Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb) MLH1, MSH2, MSH6, PMS2	4 - 6 Wo	E
Nephroblastom und Wilms-Tumor (WT) * Gen-Panel: ID335.01, 30 Gene (97,7 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CHEK2, CTR9, DICER1, DIS3L2, FBXW7, FIBP, GPC3, GPC4, KDM3B, NSD1, NYNRIN, MLH1, MSH2, MSH6, PALB2, PMS2, POU6F2, REST, TP53, TRIM28, TRIM37, TRIP13, WT1	3 - 5 Wo	E
Neuroendokrine Neoplasie * Gen-Panel: ID386.00, 19 Gene (29,2 kb) AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL	3 - 5 Wo	E
Neurofibromatose (NF) *, # Gen-Panel: ID210.00, 3 Gene (11,6 kb) NF1, NF2, SPRED1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Nierenzellkarzinom * Gen-Panel: ID041.04, 34 Gene (76,8 kb) BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1	3 - 5 Wo	E
Ovarialkarzinom * Gen-Panel: ID004.04, 14 Gene (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 Wo	E
Pankreaskarzinom * Gen-Panel: ID089.04, 19 Gene (59,9 kb) APC, ATM, BARD1, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL, WT1	3 - 5 Wo	E
Phäochromozytom-Paragangliom-Syndrom (PPGL) * Gen-Panel: ID042.03, 14 Gene (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E
Plasmozytom * Gen-Panel: ID354.01, 40 Gene (89,5 kb) ARID1A, ATM, BLM, BTK, CASP8, CASP10, CDKN2A, CBL, CTLA4, DDX41, EFL1, ETV6, FANCA, FAS, FASLG, HCLS1, KDM1A, KLHDC8B, KRAS, LAPTM5, MLH1, MSH2, MSH6, MYD88, NBN, NF1, NRAS, PAX5, PRF1, PMS2, POT1, PRKCD, PTPN11, RBM8A, SBDS, SH2B3, SH2D1A, TP53, USP45, WAS	3 - 5 Wo	E
Polyposis-Syndrom (PS, FAP) * Gen-Panel: ID005.06, 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
Prostatakarzinom * Gen-Panel: ID140.03, 30 Gene (101,5 kb) AR, ATM, ATR, BAP1, BARD1, BRCA1, BRCA2, BRIP1, CYP3A43, CDH1, CHEK2, ELAC2, EPCAM, FANCM, HOXB13, MLH1, MRE11, MSR1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, RNASEL, TP53, TRRAP	3 - 5 Wo	E
Sarkom der Weichteile und des Skelettsystems * Gen-Panel: ID223.02, 55 Gene (155,1 kb) APC, ATM, ATR, ATRX, BLM, BRCA2, BUB1B, CDKN1C, CDKN2A, CEP57, CHEK2, DICER1, DKC1, EPCAM, ERCC2, EXT1, EXT2, FAH, FANCC, FH, HRAS, KIT, MEN1, MLH1, MRE11, MSH2, MSH6, MTAP, NBN, NF1, PALB2, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RPS19, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SQSTM1, SUFU, TBXT, TNFRSF11A, TP53, VHL, WAS, WRN, ZNF687	4 - 6 Wo	E
Schilddrüsenkarzinom * Gen-Panel: ID220.02, 26 Gene (60,5 kb) ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53	3 - 5 Wo	E
Tuberöse Sklerose (TSC) * Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
Urothelkarzinom * Gen-Panel: ID337.00, 34 Gene (105,0 kb) APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC	3 - 5 Wo	E
Xeroderma pigmentosum (XP) * Gen-Panel: ID282.00, 10 Gene (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 Wo	E
Ziliopathien		
Bardet-Biedl-Syndrom (BBS) * Gen-Panel: ID093.02, 21 Gene (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDRCP	3 - 5 Wo	E
Joubert-Syndrom (JBTS) * Gen-Panel: ID028.03, 40 Gene (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 Wo	E
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD) * Gen-Panel: ID067.00 Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD): 20 Gene (68,2 kb) CEP120, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Kurzrippen-Thoraxdysplasie (Jeune-ATD, SRPS, SRTD): 18 Gene (61,3 kb) CEP120, DYNC2H1, DYNC2LI1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Ellis-van-Crefeld-Syndrom (EVC): 4 Gene (11,5 kb) DYNC2LI1, EVC, EVC1, WDR35	3 - 5 Wo	E
Meckel-Syndrom (MKS) * Gen-Panel: ID032.02, 13 Gene (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 Wo	E
Nephronophthise (NPHP) * Gen-Panel: ID030.02, 22 Gene (70,9 kb) ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Ziliopathien		
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) * Gen-Panel: ID085.03, 50 Gene (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 Wo	E
Senior-Loken-Syndrom (SLSN) * Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E
Renale Ziliopathien, umfassende Diagnostik * Gen-Panel: ID376.00 Renale Ziliopathien, umfassende Diagnostik: 75 Gene (181,7 kb) AHI1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPGRIP1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, XPNPEP3, ZNF423 Nephronophthise (NPHP): 17 Gene (50,3 kb) ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP Senior-Loken-Syndrom (SLSN): 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 Joubert-Syndrom (JBTS): 40 Gene (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423 Meckel-Syndrom (MKS): 14 Gene (36,2 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15	4 - 6 Wo	E
Präventionsdiagnostik: Carrier-Screening		
Carrier-Screening * Gen-Panel: ID350.00, 625 Gene (1591,4 kb) AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACOX1, ACSF3, ADA, ADAMTS13, ADAMTS2, ADGRG1, ADGRV1, AFF2, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMT, ANK1, ANO10, ANTXR2, APTX, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6VOA2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRIP1, BSND, BTD, BTK, CA2, CAPN3, CASR, CBS, CC2D2A, CCDC88C, CD3D, CD3E, CD40LG, CDH23, CEP290, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNG, CIITA, CLCN1, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL1A2, COL27A1, COL4A3, COL4A4, COL4A5, COL7A1, COQ2, COQ8A, CPS1, CPT1A, CPT2, CRB1, CRLF1, CRTAP, CSTB, CTNS, CTSC, CTSD, CTSK, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP27A1, CYP27B1, D2HGDH, DBT, DCLRE1C, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DHDDS, DKC1, DLD, DLL3, DMD, DMP1, DNAH5, DNAI1, DNAI2, DNAJC19, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPYD, DSP, DUOX2, DUOX2A, DYNC2H1, DYSF, EDA, EDN3, EDNRB, EFEMP2, EGR2, EIF2AK3, EIF2B5, ELP1, EMD, ENPP1, EPB42, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, F11, F8, F9, FAX, FAM126A, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCG, FBLN5, FBP1, FGA, FGB, FGD4, FGG, FH, FKRP, FKTN, FLNA, FMO3, FOLR1, FOXP1, FOXP3, FRAS1, FREM2, FUCA1, FXN, G6PC1, G6PC3, G6PD, GAA, GALT, GALE, GALK1, GALNS, GALT, GAMT, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDF5, GFM1, GJA1, GJB1, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNE, GNPTAB, GNPTG, GNS, GP1BA, GP9, GRHPR, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HESX1, HEXA, HEXB, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HOGA1, HPD, HPRT1, HPS1, HPS3, HPS4, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPG2, HYAL1, HYLS1, IDS, IDUA, IGBP1, IGF1, IGHMBP2, IGSF1, IL2RG, IL7R, INPP5E, INS, INSR, INVS, IQCB1, ITGA6, ITGB4, IVD, IYD, JAG1, JAK3, KCNJ1, KCNJ11, KCNQ1, KCNQ2, KCTD7, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LANGE1, LBR, LCA5, LDLR, LDLRAP1, LHCRG, LHX3, LIFR, LIPA, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MED12, MED17, MEFV, MESP2, MFSD8, MGAT2, MID1, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPI, MPL, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTPP, MUTHY, MYK, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFAF5, NDUFS6, NEU1, NEUROG3, NHLRC1, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NROB1, NR2E3, NTRK1, NUP62, OAT, OCA2, OCLR, OFD1, OPA3, OSTM1, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRCT, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLRC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRCT, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A20, SLC26A2, SLC26A3, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A1, SLC4A11, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMARCAL1, SMPD1, SNAP29, SP110, SPR, SRD5A2, ST3GAL5, STAR, STRA6, SUGL1, SUMF1, SUOX, TAFAZZIN, TAT, TBCE, TCIRG1, TCEPR2, TF, TFR2, TG, TGM1, TH, TIMM8A, TK2, TMEM216, TMEM67, TRNRSF11B, TNNI1, TPO, TPP1, TREX1, TRHR, TRIM32, TRIM37, TRMU, TSEN54, TSHB, TSHR, TSPYL1, TTC37, TTN, TTPA, TWNK, TYMP, TYR, TYRP1, UBA1, UBR1, UGT1A1, UQCRB, UQCQR, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WAS, WNT10A, WNT3, WNT7A, WRN, WT1, XPA, XPC, ZIC3, ZMPSTE24, ZNF469	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Pränatale Diagnostik: Fetale Anomalien		
Fetale Anomalien * 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, AMER1A, AMMECR1, AMPD2, AMT, ANAPC1, ANKH, ANKRD11, ANKS6, ANOS1, ANTXR1, ANTXR2, AP1S2, AP4E1, AR, ARCN1, ARFGF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMCD, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6VOA2, 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, BMPEP, BMPR1B, BNC2, BPNT2, BRAF, BRAT1, BRCA2, BRIP1, BRPF1, BSND, BTBD, 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, CHRNA3, CHRNBP, CRTAP, CRLF1, CRPPA, CRTAP, CRYAA, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CSF1R, CSNK2A1, CSPP1, CTC1, CTCF, CTNNB1, CTNNB1, CTNSA, CTSD, CTSK, CTSP, CUL4B, CUL7, CWC27, CYP11A1, CYP11B1, CYP17A1, CYP1B1, CYP21A2, CYP26B1, CYP2U1, CYP4F22, DAG1, DARS1, DCHS1, DCX, DDR2, DDX11, DDX3X, DDX59, DENND5A, DHCR24, DHCR7, DHFR, DHODH, DIAPH1, DIS3L2, DISP1, DKC1, DLL3, DLL4, DLX5, DMFP, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAIL2, DNABJ11, DNAL1, DNML1, DNMT2, DNMT3B, DNMT3B, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSP, DSTYK, DVLL1, IDH1, DYM, DYNC1H1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, DYRK1A, DZIP1L, EBF3, EBP, ECEL1, EDA, EDNRA, EDNRB, EED, EFNB1, EFTUD2, EHMT1, EIF2AK3, EIF2B2, EIF2B3, EIF2S3, EIF4A3, EIF5A, ELAC2, ELN, ELOVL4, EMD, EML1, EMX2, ENPP1, EOBT, EP300, EPG5, EPHB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERCC8, ERF, ESCO2, ETFA, ETFB, ETFDH, EVC, EVD, EXOC3L2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EZH2, FAH, FAM111A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANGC, FANCI, FANCL, FAR1, FAT4, FBLN5, FBN1, FBN2, FBXL4, FGDI, FGF10, FGF3, FGF8, FGFR1, FGFR2, FGFR3, FH, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FLCN, FLT4, FLVCR2, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMD4A, FTL, FUT8, FYCO1, FZD2, G6PC3, GAA, GALT, GALK1, GALN3, GALNT2, GANAB, GATA2, GATA3, GATA4, GATA6, GBA1, GBA2, GBEL, GCDH, GDF1, GDF5, GDF6, GFAP, GFM1, GFFT1, GJA1, GJA3, GJB8, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLI1, GLI2, GLI3, GLIS3, GLUL, GMNN, GMPBP, GNAI3, GNAO1, GNAS, GNB1, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC6, GPI, GPSM2, GREB1L, GRHL3, GRIN1, GRIN2B, GRIP1, GSC, GTFH25, GTPBP3, GUCY2C, GUSB, GZF1, H1-4, HAAO, HADHA, HADHB, HBA1, HBA2, HCOS, HCF1, HDAC8, HESE7, HESX1, HIBCH, HIVEP2, HMGA2, HNFB1B, HNFA4, HNRNP, HOXA1, HOXA13, HOXD13, HPSE2, HR, HRAS, HSD17B3, HSD17B4, HSFA, HSPD1, HSPG2, HUWE1, HYCC1, HYL1, IERS1, IDH1, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGF2, IGHMBP2, IHH, IKBKG, IL11RA, IL1RAPL1, INPP5E, INPPL1, INSR, INTU, INVS, IQCB1, IRF6, ITGA3, ITGA6, ITGA8, ITGB4, JAG1, KANS1, KAT6A, KAT6B, KATNB1, KCNJ1, KCNJ2, KCTD1, KDM5C, KDM6A, KIAA0586, KIAA0753, KIF11, KIF14, KIF1A, KIF1B, KIF22, KIF2A, KIF5C, KIF7, KLF1, KLHL40, KLHL41, KLHL7, KMT2A, KMT2C, KMT2D, KNL1, KRAS, KRIT1, KYNU, L1CAM, LZHGHD, LAMA1, LAMA2, LAMB1, LAMC3, LARG1, LARP7, LBR, LFNG, LGI4, LHX4, LHXA, LIFR, LIG4, LIPA, LMBR1, LMBRD1, LMNA, LMNB1, LMNB2, LMOD3, LMX1B, LONP1, LRPI, LRP2, LRP4, LRP5, LRRC56, LTBP3, LTBP4, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAGEL2, MAP2K1, MAP2K2, MAP3K1, MAP3K20, MAP3K7, MAPRE2, MASP1, MATN3, MBTPS2, MCOLN1, MCPH1, MED12, MEFE2C, MEGF10, MEGF8, MEIS2, MEOX1, MESD, MESPD2, MFRR, MFSFD2A, MG, MID1, MKKS, MKS1, MLC1, MLFY, MMACHC, MMADHC, MMP13, MMP21, MN1, MNX1, MOCS1, MOCS2, MOGS, MPDU1, MPLKIP, MRAS, MRPS22, MSL3, MSMO1, MSTO1, MSX1, MSX2, MTM1, MTO1, MTOR, MTRFR, MUSK, MYBPC1, MYCN, MYH10, MYH11, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL1, MYMK, MYO18B, MYO9A, MYOCD, MYPN, MYRF, MYT1, NAA10, NACC1, NADSYN1, NAGA, NALCN, NANS, NBAS, NBN, NDE1, NDP, NDUF4F5, NEB, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEK9, NEU1, NYF1, NFIX, NHEJ1, NHS, NIPAL4, NIPBL, NKX2-5, NKX3-2, NOTAL, NOG, NOTCH1, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPR2, NROB1, NR2F2, NR5A1, NRAS, NSD1, 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RAB3GAP2, RAC1, RAD21, RAF1, RA1, RAPSN, RARB, RARS2, RASA1, RAX, RBBP8, RBM10, RBM8A, RBPJ, REQLQ4, RELN, REN, RERE, RET, RFT1, RFKB, RFX4, RFX1, RFXP4, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU4ATAC, ROBO1, ROBO3, ROGDI, ROR2, RPGRIP1L, RPL10, RPL11, RPL35A, RPL5, RPLK1, RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7, RRAS2, RRM2B, RSHP4A, RSHP9, RTDL1, RTTN, RUNX2, RXLYT1, RYR1, SALL1, SALL4, SAMD9, SAMHD1, SATB2, SBDS, SC5D, SCARF2, SCLT1, SCN1A, SCN2A, SCN4A, SCO2, SDCCAG8, SDR9C7, SEC23B, SEC24D, SEPSEC5, SERPINF1, SERPINH1, SETBP1, SETD5, SF3B4, SGCG, SGP1L, SH3PXD2B, SHANK3, SHH, SHOC2, SHOX, SIK3, SIL1, SIX3, SIX5, SIX6, SKI, SKIC3, SLC10A7, SLC12A1, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC18A3, SLC25A19, SLC25A20, SLC25A24, SLC25A38, SLC26A4, SLC26A3, SLC27A4, SLC29A3, SLC32A1, SLC35A2, SLC35A1, SLC35C1, SLC35D1, SLC39A8, SLC5A7, SLC6A9, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC1, SMARCE1, SMC1A, SMC3, SMCHD1, SMG9, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNORD118, SNRPB, SNX10, SNX14, SON, SOS1, SOS2, SOST, SOX10, SOX17, SOX18, SOX2, SOX3, SOX6, SOX9, SP7, SPAG1, SPARC, SPATAS, SPECC1L, SPEG, SPG11, SPG2, SPG12, SRPAC, SRD5A2, SRD5A3, SRY, ST14, STAC3, STAG2, STAMBP, STAR, STIL, STRA6, STRADA, SUCLG1, SUFU, SULT2B1, SUMF1, SUZ12, TAB2, TAF1, TAFAZZIN, TALDO1, TAPT1, TBC1D20, TBC1D23, TBC1D24, TBC1D32, TBCD, TBCE, TBCK, TBL1XR1, TBX1, TBX15, TBX18, TBX20, TBX3, TBX4, TBX5, TBX6, TCF12, TCF4, TCIRG1, TCOF1, TCTN1, TCTN2, TCTN3, TLO2, TENM3, TENT5A, TFAP2A, TFAP2B, TGDS, TGF2, TGF3, TGFBR1, TGFBR2, TGIF1, TGM1, THOC6, THRA, TINF2, TMC01, TMC02, TMEM107, TMEM138, TMEM165, TMEM216, TMEM237, TMEM388, TMEM67, TMEM94, TMEM98, TMX2, TNNI2, TNNT1, TNNT3, TOE1, TOP3A, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRAF7, TRAI, TRAP1, TRAPPC12, TRAPPC9, TREX1, TRIM37, TRIP11, TRIP12, TRIP4, TRMT10A, TRPS1, TRPV4, TRPV6, TSC1, TSC2, TSEN2, TSEN3A, TSEN54, TSMF, TTC21B, TTC7A, TTC8, TTN, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TWIST2, TXNDC15, TXNL4A, UBA1, UBE2T, UBE3B, UBR1, UMP, UROS, USP18, USP9X, VAMP1, VEGFC, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDCP, WDR19, WDR26, WDR35, WDR62, WDR73, WDR81, WNT1, WNT10B, WNT5A, WNT7A, WRAP53, WT1, XRC4, XYL1, XYL2, YY1, ZBTB18, ZBTB20, ZC4H2, ZEB2, ZFP57, ZIC1, ZIC2, ZIC3, ZMPSTE24, ZMYND10, ZSWIM6	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
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		