SYNLAB Humangenetik

Stand: 12/2023

Next Generation Sequencing (NGS)

Panel-Verzeichnis

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Achromatopsie (ACHM) * Gen-Panel: ID164.01, 7 Gene (11,5 kb) ATF6, CNGA3, CNGB3, GNAT2, OPN1LW, 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
Dysgenesie des vorderen Augensegmentes (ASGD) * Gen-Panel: ID182.02 Dysgenesie des vorderen Augensegmentes (ASGD): 9 Gene (18,8 kb) B3GLCT, CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN Peters-Anomalie: 6 Gene (7,8 kb) B3GLCT, CYP1B1, FOXC1, PAX6, PITX2, PITX3 Axenfeld-Rieger-Anomalie: 2 Gene (3,3 kb) FOXC1, PITX2	3 - 5 Wo	E
Fraser-Syndrom (FRASRS) * Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
Fuchs-Endotheldystrophie (FECD) * Gen-Panel: ID261.00, 5 Gene (13,5 kb) AGBL1, COL8A2, SLC4A11, TCF4, ZEB1	3 - 5 Wo	E
Glaukom (GLC) * Gen-Panel: ID275.01 Glaukom (GLC): 27 Gene (53,5 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CPAMD8, CYP1B1, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, PAX6, PITX2, PITX3, PXDN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMCO1, WDR36 Primäres Glaukom (GLC1, GLC3): 20 Gene (37,8 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CYP1B1, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMCO1, WDR36 Dysgenesie des vorderen Augensegmentes (ASGD): 8 Gene (17,5 kb) CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS) * Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
Hornhautdystrophie (CD) * Gen-Panel: ID329.00 Hornhautdystrophie (CD): 26 Gene (67,0 kb) AGBL1, CHST6, COL8A2, COL17A1, CYP4V2, DCN, FOXE3, GRHL2, GSN, KRT3, KRT12, LCAT, LOXHD1, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469 Fuchs-Endotheldystrophie (FECD): 6 Gene (20,2 kb) AGBL1, COL8A2, LOXHD1, SLC4A11, TCF4, ZEB1 Hintere polymorphe Hornhautdystrophie (PPCD): 4 Gene (8,2 kb) COL8A2, GRHL2, OVOL2, ZEB1 Gittrige Hornhautdystrophie (CDL): 3 Gene (5,4 kb) GSN, TACSTD2, TGFBI Stromale Hornhautdystrophie: 5 Gene (11,6 kb) CHST6, DCN, PIKFYVE, TGFBI, UBIAD1 Epitheliale Hornhautdystrophie: 5 Gene (10,9 kb) COL17A1, KRT3, KRT12, TACSTD2, TGFBI Brittle-Cornea-Syndrom (BCN): 2 Gene (13,8 kb) PRDM5, ZNF468 Keratokonus (KTCN): 2 Gene (2,5 kb) TUBA3D, VSX1	3 - 5 Wo	E
Kongenitale extraokuläre Muskelfibrose (CFEOM) * Gen-Panel: ID063.00, 5 Gene (10,5 kb) COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3	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

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
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
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
Mikrophthalmie, Anophthalmie und Kolobom * Gen-Panel: ID263.01 Mikrophthalmie, Anophthalmie und Kolobom: 50 Gene (120,8 kb) ABCB6, ALDH1A3, BCOR, BMP4, CC2D2A, CHD7, C12ORF57, CRYAA, FRAS1, FREM1, FREM2, FOXE3, GDF3, GDF6, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MAF, MFRP, MITF, NAA10, OTX2, PAX2, PAX6, PDE6D, PIGL, PITX3, PTCH1, PRSS56, RARB, RAX, RBP4, RPGRIP1L, SALL2, SEMA3A, SHH, SIX3, SMOC1, SOX2, SRD5A3, STRA6, TENM3, TFAP2A, TMEM67, VAX1, VSX2, YAP1 Mikrophthalmie, syndromal (MCOPS): 12 Gene (23,9 kb) BCOR, BMP4, HCCS, HMGB3, MAB21L2, NAA10, OTX2, SOX2*, RARB, STRA6, TENM3, VAX1 Mikrophthalmie, isoliert (MCOP): 7 Gene (9,7 kb) ALDH1A3, GDF3, GDF6, MFRP, PRSS56, RAX, VSX2 Mikrophthalmie, isoliert, mit Kolobom (MCOPCB): 8 Gene (18,2 kb) ABCB6, GDF3, GDF6, RBP4, SHH, STRA6, TENM3, VSX2 Okulares Kolobom, isoliert: 2 Gene (4,3 kb) PAX6, SALL2	4 - 6 Wo	E
Morbus Stargardt (STGD) * Gen-Panel: ID102.00, 5 Gene (13,8 kb) ABCA4, CNGB3, ELOVL4, PROM1, PRPH2	3 - 5 Wo	E
Myopie (MYP) * Gen-Panel: ID079.02 Myopie (MYP): 20 Gene (61,1 kb) ARR3, CNGB3, CPSF1, COL11A1, COL2A1, COL9A1, COL9A2, COL18A1, GZF1, IRX5, LRPAP1, LRP2, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644 Myopie (MYP), nicht-syndromal: 11 Gene (22,3 kb) ARR3, CNGB3, CPSF1, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644 Myopie (MYP), syndromal: 10 Gene (41,0 kb) COL11A1, COL2A1, COL9A1, COL9A2, COL18A1, GZF1, LRP2, IRX5, P3H2, SLITRK6 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL11A1, COL2A1, COL9A1, COL9A	3 - 5 Wo	E
Nystagmus (NYS) * Gen-Panel ID331.00 Nystagmus (NYS): 44 Gene (105,8 kb) ATF6, CACNA1A, CACNA1F, CASK, CEP290, CNGA3, CNGB3, CRB1, CRX, DCT, FRMD7, GJC2, GNAT2, GPR143, GUCY2D, HPS1, HPS5, IMPDH1, KCNJ13, KIDINS220, LCA5, LRAT, LRMDA, LYST, MAG, NMNAT1, NYX, OCA2, OPN1LW, OPN1MW, PAX6, PDE6C, PDE6H, PLP1, PRPH2, RPE65, RPGRIP1, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1, USP45 Nystagmus, nicht-syndromal (NYS): 2 Gene (3,4 kb) FRMD7, GPR143 Foveahypoplasie, nicht-syndromal (FVH): 2 Gene (2,6 kb) PAX6, SLC38A8 Albinismus (OCA): 11 Gene (29,2 kb) DCT, GPR143, HPS1, HPS5, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Achromatopsie (ACHM): 8 Gene (12,6 kb) ATF6, CNGA3, CNGB3, GNAT2, OPN1LW, OPN1MW, PDE6C, PDE6H Lebersche kongenitale Amaurose (LCA): 14 Gene (33,0 kb) CEP290, CRB1, CRX, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RPE65, RPGRIP1, TULP1, USP45 Pelizaeus-Merzbacher-Krankheit (PMD): 3 Gene (4,0 kb) PLP1, GJC2, MAG	4 - 6 Wo	E
Okulokutaner Albinismus (OCA) * Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Optikusatrophie (OPA) * Gen-Panel ID081.04 Optikusatrophie (OPA): 36 Gene (57,9 kb) ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, 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: 10 Gene (14,8 kb) ACO2, AFG3L2, DNM1L, OPA1, OPA3, RTN4IP1, SPG7, SSBP1, TMEM126A, YME1L1 Optikusatrophie (OPA), syndromal: 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
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: ID053.02, 25 Gene (54,6 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RGR, RHO, RP1, RP9, RPE65, SEMA4A, SNRNP200, TOPORS	3 - 5 Wo	E
Retinitis pigmentosa (RP), autosomal-rezessiv * Gen-Panel: ID050.03, 57 Gene (153,2 kb) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, C8ORF37, CDHR1, CERKL, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513	4 - 6 Wo	E
Retinitis pigmentosa (RP), umfassende Diagnostik * Gen-Panel: ID288.02 Retinintis pigmentosa (RP), umfassende Diagnostik: 87 Gene (216,9 kb) ABCA4, AGBL5, AHR, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS2, BEST1, CC2D2A, CDHR1, CERKL, CFAP418, CHM, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, DHX38, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, HGSNAT, HK1, HKDC1, IDH3A, IDH3B, IFT140, IFT172, IFT43, IMPDH1, IMPG1, IMPG2, KIAA1549, KIF3B, KIZ, KLHL7, LRAT, MAK, MERTK, NEK2, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF408, ZNF513 Retinitis pigmentosa (RP), autosomal-dominant: 28 Gene (59,2 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPDH1, IMPG1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS Retinitis pigmentosa (RP), autosomal-rezessiv: 61 Gene (162,4 kb) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513 Retinitis pigmentosa (RP), X-chromosomal: 4 Gene (8,5 kb) CHM, RP2, RPGR, OFD1	4 - 6 Wo	E
Senior-Loken-Syndrom (SLSN) * Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 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
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

Erkrankung/Diagnostik	Dauer	Material
Augenerkrankungen		
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Weill-Marchesani-Syndrom (WMS) * Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD) * Gen-Panel: ID101.02 Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD): 29 Gene (72,4 kb) ABCA4, ADAM9, AIPL1, C8ORF37, CACNA1F, CACNA2D4, CDHR1, CEP78, CEP250, CNNM4, CRX, DRAM2, GUCA1A, GUCY2D, KCNV2, OPN1LW, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5, UNC119 Zapfen-Stäbchen-Dystrophie (CORD): 24 Gene (63,4 kb) ABCA4, ADAM9, AIPL1, C8ORF37, CACNA1F, CDHR1, CEP78, CEP250, CNNM4, CRX, DRAM2, GUCA1A, GUCY2D, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5, UNC119 Zapfen-Dystrophie (COD): 8 Gene (15,3 kb) CACNA2D4, GUCA1A, GUCY2D, KCNV2, OPN1LW, PDE6C, PDE6H, RPGR	3 - 5 Wo	E
Bindegewebserkrankungen		
Bindegewebserkrankungen mit Aortenbeteiligung * Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, TNXB, ZNF469 Thorakales Aortenaneurysma und Aortendissektion (TAA/D): 17 Gene (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFBR2, TGFBR1 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
Ehlers-Danlos-Syndrom (EDS) * Gen-Panel: ID039.05 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos-Syndrom (EDS), autosomal-dominant: 8 Gene (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 , COL12A1 Ehlers-Danlos-Syndrom (EDS), autosomal-rezessiv: 13 Gene (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	3 - 5 Wo	E
Hereditäre hämorrhagische Teleangiektasie (HHT) * Gen-Panel: ID155.01, 7 Gene (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 Wo	E
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E
Marfan-Syndrom (MFS) *, # Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1 TGFBR1, TGFBR2	2 - 4 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Bindegewebserkrankungen		
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.03 Marfan-Syndrom und ähnliche Krankheitsbilder: 39 Gene (134,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL11A1, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MTHFR, MYH11, MYLK, PLOD1, PRDM5, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 Thorakales Aortenaneurysma (TAA/D): 13 Gene (36,4 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MYH11, MYLK, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469 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
Osteogenesis imperfecta (OI) * Gen-Panel: ID066.02, 21 Gene (39,2 kb) ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1	3 - 5 Wo	E
Stickler-Syndrom (STL) * Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) * Gen-Panel: ID020.01 Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 16 Gene (41,0 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	3 - 5 Wo	E
Endokrine Störungen		
Adrenogenitales Syndrom (AGS, CAH) * Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
Diabetes insipidus * Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E
Endokrine Hypertonie * Gen-Panel: ID270.02 Endokrine Hypertonie: 32 Gene (80,9 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 6 Gene (20,6 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom/Paragangliom-Syndrom (PPGL): 9 Gene (9,5 kb) MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, 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	3 - 5 Wo	E
Glukokortikoid-Mangel (GCCD) * Gen-Panel: ID222.00, 16 Gene (25,1 kb) AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NR0B1, NR3C1, PCSK1, POMC, PROP1, STAR, TBX19, TXNRD2	3 - 5 Wo	E
Hyperaldosteronismus (HALD) * Gen-Panel: ID304.00, 6 Gene (20,6 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5	3 - 5 Wo	E
Hyperinsulinämische Hypoglykämie (HHF) * Gen-Panel: ID126.00, 8 Gene (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 Wo	E
Hyperparathyreoidismus (HRPT) * Gen-Panel: ID338.00, 10 Gene (19,2 kb) AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
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: ID170.04, 37 Gene (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, 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
Kongenitale Hypothyreose * Gen-Panel: ID369.00 Kongenitale Hypothyreose: 37 Gene (69,0 kb) CDCA8, DIO1, DUOX2, DUOXA2, FOXE1, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, NKX2-1, NKX2-5, OTX2, PAX8, POU1F1, PRKAR1A, PROP1, RNPC3, ROBO1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TUBB1 Kongenitale Schilddrüsenunterfunktion ohne Kropf (CHNG): 8 Gene (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Schilddrüsen-Dyshormonogenese (TDH): 8 Gene (23,9 kb) DUOX2, DUOXA2, IYD, SLC5A5, SLC26A4, SLC26A7, TG, TPO Kombinierter Hypophysenhormonmangel (CPHD): 8 Gene (11,9 kb) HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1	3 - 5 w	E
MODY-Diabetes * Gen-Panel: ID048.01, 14 Gene (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 Wo	E
Neonataler Diabetes mellitus * Panel: ID162.01 Neonataler Diabetes mellitus: 29 Gene (53,6 kb) ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57 Permanenter neonataler Diabetes mellitus (PNDM): 10 Gene (15,9 kb) ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57 Syndromaler neonataler Diabetes mellitus: 21 Gene (40,5 kb) BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 Wo	E
Ovarialdysgenesie (ODG) * Gen-Panel: ID293.01 Ovarialdysgenesie (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX-Gonadendysgenesie (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX-Gonadendysgenesie mit Taubheit (PRLTS): 6 Gene (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 Wo	E
Störung der Geschlechtsentwicklung (DSD) * Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSPO1, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
Paragangliom und Phäochromozytom * Gen-Panel: ID042.02, 16 Gene (29,4 kb) DLST, FH, KIF1B, GDNF, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Endokrine Störungen		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) *	3 - 5 Wo	E
Gen-Panel: ID250.01		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PAH): 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		
Vorzeitige Ovarialinsuffizienz (POF) *	3 - 5 Wo	E
Gen-Panel: ID078.05		
Vorzeitige Ovarialinsuffizienz (POF): 23 Gene (51,1 kb)		
BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, INHA, LHCGR, MCM8, MSH5, NOBOX, NR5A1, POF1B, STAG3, SYCE1, XRCC2		
Vorzeitige Ovarialinsuffizienz (POF), X-chromosomal: 4 Gene (8,2 kb)		
BMP15, DIAPH2, FMR1, POF1B		
Vorzeitige Ovarialinsuffizienz (POF), autosomal-rezessiv: 10 Gene (25,0 kb)		
C14ORF39, GDF9, FANCM, HFM1, HSF2BP, MCM8, MSH5, STAG3, SYCE1, XRCC2		
Vorzeitige Ovarialinsuffizienz (POF), autosomal-dominant: 6 Gene (12,7 kb)		
BNC1, ERCC6, FIGLA, FOXL2, NOBOX, NR5A1		
Wachstumshormonmangel (IGHD, CPHD) *	3 - 5 Wo	E
Gen-Panel: ID211.02		
Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb)		
BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNPC3, OTX2, POU1F1, PROP1, SEMA3A, SOX3		
Isolierter Wachstumshormonmangel (IGHD): 5 Gene (6,4 kb)		
BTK, GH1, GHRHR, GHSR, SOX3		
Kombinierter Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb)		
GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A		
Entwicklungs- und Wachstumsstörungen		
Autismus-Spektrum-Störungen *	4 - 6 Wo	E
Gen-Panel: ID076.03		
Autismus-Spektrum-Störungen: 106 Gene (377,1 kb)		
ADNP, ADSL, AFF2, ALDH5A1, ARX, ARID1B, ASH1L, ASTN2, ATP1A1, AUTS2, BCL11A, CACNA1C, CASK, CC2D1A, CDKL5, CHD2, CHD7, CHD8, CNOT3, CNTN4, CNTNAP2, CTNNB1, CUL3, DEAF1, DHCR7, DLGAP2, DPP6, DYRK1A, EHMT1, EIF4E, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, GRIA3, GRIN2A, GRIN2B, HERC2, IL1RAPL1, IQSEC2, KDM5C, KMT2C, KTM5B, MAOA, MBD5, MECP2, MED12, MED13L, MEF2C, MYT1L, NAA15, NAGLU, NEXMIF, NHS, NLGN1, NLGN3, NLGN4X, NR1I3, NRXN1, NRXN2, NRXN3, OPHN1, PAH, PCDH19, PHF6, PHF8, PHF21A, POGZ, PQBP1, PTCHD1, PTEN, RAB39B, RAI1, RPL10, SCN1A, SCN2A, SEMA5A, SETD2, SGSH, SHANK2, SHANK3, SLC6A1, SLC6A8, SLC9A6, SLC9A9, SLC35A3, SMARCB1, SYN1, SYNGAP1, TBL1XR1, TBR1, TCF4, TLK2, TMLHE, TRRAP, TSC1, TSC2, UBE2A, UBE3A, UPF3B, VPS13B, ZEB2, ZIC1, ZMYND11, ZNF292		
Prädisposition für Autismus (AUTS): 14 Gene (36,5 kb)		
CHD8, CNTNAP2, EIF4E, MECP2, NLGN1, NLGN3, NLGN4X, PTCHD1, PTEN, RPL10, SHANK2, SLC9A9, TBR1, TMLHE		
Coffin-Siris-Syndrom (CSS) *	3 - 5 Wo	E
Gen-Panel: ID118.01, 10 Gene (34,2 kb)		
ARID1A, ARID1B, ARID2, DPF2, SMARCC2, SMARCA4, SMARCB1, SMARCE1, SOX4, SOX11		
Cornelia-de-Lange-Syndrom (CDLS) *	3 - 5 Wo	E
Gen-Panel: ID033.02, 8 Gene (32,0 kb)		
ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS		
FG-Syndrom (FGS) *	3 - 5 Wo	E
Gen-Panel: ID215.00, 3 Gene (17,2 kb)		
CASK, FLNA, MED12		
Großwuchs, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID299.01, 64 Gene (211,5 kb)		
ABCC9, AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, FBN1, FIBP, GFAP, GLI3, GPC3, GPC4, GRIA3, H1-4, HEPACAM, HERC1, HUWE1, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIGA, PIK3CA, PIK3R2, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SHANK, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMEM94, TRIP12, UPF3B, ZBTB20, ZDHHC9		
Großwuchssyndrome (SOTOS, BWS) *	3 - 5 Wo	E
Gen-Panel: ID073.05		
Großwuchssyndrome (SOTOS, BWS): 15 Gene (41,2 kb)		
CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, PDGFRB, PTEN, SETD2, SUZ12, RNF125		
Sotos-ähnliche Großwuchssyndrome: 6 Gene (23,1 kb)		
EED, EZH2, NFIX, NSD1, SETD2, SUZ12		
Beckwith-Wiedemann-ähnliche Großwuchssyndrome: 5 Gene (10,1 kb)		
CDKN1C, DIS3L2, GPC3, GPC4, OFD1		
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *	3 - 5 Wo	E
Gen-Panel: ID292.00, 6 Gene (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) *	4 - 6 Wo	E
Gen-Panel: ID036.05		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD): 74 Gene (258,2 kb)		
ADNP, AHDC1, AP2M1, ARID1A, ARID1B, ARID2, ASH1L, ATP2B1, AUTS2, BICRA, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CHAMP1, CIC, CLTC, CTCF, CTNNB1, DEAF1, DLG4, DPF2, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN1, GRIN2B, HIVEP2, KAT6A, KCNQ5, KDM4B, KIF1A, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MEF2C, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, PURA, RAC1, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMARCD1, SOX4, SOX11, STAG1, SYNGAP1, TBL1XR1, TLK2, TRIO, TRIP12, ZBTB18, ZMYND11, ZNF292		
Intellektuelle Entwicklungsstörung, autosomal-dominant, nicht-syndromal (MRD): 57 Gene (203,8 kb)		
AP2M1, ARID1A, ARID1B, ASH1L, ATP2B1, AUTS2, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CIC, CLTC, CTCF, DEAF1, DLG4, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN2B, HIVEP2, KCNQ5, KDM4B, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, RAC1, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SOX11, STAG1, SYNGAP1, TBL1XR1, TLK2, TRIO, ZBTB18, ZMYND11, ZNF292		
Intellektuelle Entwicklungsstörung, autosomal-dominant, syndromal: 26 Gene (86,4 kb)		
ADNP, AHDC1, ARID1A, ARID1B, ARID2, BICRA, CHAMP1, CTNNB1, DEAF1, DPF2, GATAD2B, GRIN1, KAT6A, KIF1A, MEF2C, PACS1, POGZ, PURA, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMARCD1, SOX4, SOX11, TRIP12		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) *	4 - 6 Wo	E
Gen-Panel: ID037.02, 54 Gene (141,2 kb)		
ADAT3, ALKBH8, ANK3, APC2, C12ORF4, CAMK2A, CC2D1A, CEP104, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31, FMN2, GRIA1, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NSUN2, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, ZBTB11, ZC3H14		
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS) *	4- 6 Wo	E
Gen-Panel: ID038.05		
Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS): 72 Gene (178,2 kb)		
ACSL4, AFF2, AP1S2, ARX, ATP6AP2, ATRX, BRWD3, CASK, CLCN4, CLIC2, CNKSR2, CUL4B, DDX3X, DLG3, EIF2S3, FAM50A, FGD1, FGF13, FMR1, FRMPD4, FTSJ1, GDI1, GLRA2, GRIA3, HCFC1, HNRNPH2, HS6ST2, HUWE1, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIF4A, KLHL15, LAS1L, MECP2, MED12, MID2, MSL3, NEXMIF, NKAP, NONO, OGT, OPHN1, PAK3, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, RPS6KA3, SLC16A2, SLC9A6, SLC9A7, SMS, STEEP1, SYN1, SYP, TAF1, THOC2, TSPAN7, UBE2A, UPF3B, USP9X, USP27X, ZC4H2, ZDHHC9, 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): 47 Gene (114,4 kb)		
AFF2, AP1S2, ARX, ATP6AP2, ATRX, CASK, CLCN4, CLIC2, CNKSR2, 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		
Intellektuelle Entwicklungsstörung und Makrozephalie *	4 - 6 Wo	E
Gen-Panel: ID131.03		
Intellektuelle Entwicklungsstörung und Makrozephalie: 48 Gene (147,3 kb)		
ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Makrozephalie: 21 Gene (70,8 kb)		
CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Makrozephalie: 10 Gene (20,2 kb)		
ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMCO1, ZBTB7A		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Makrozephalie: 18 Gene (57,9 kb)		
BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9		
Intellektuelle Entwicklungsstörung und Mikrozephalie *	4 - 6 Wo	E
Gen-Panel: ID129.02		
Intellektuelle Entwicklungsstörung und Mikrozephalie: 80 Gene (229,8 kb)		
ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNNB1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNPH2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Mikrozephalie: 32 Gene (100,5 kb)		
AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2		
Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Mikrozephalie: 21 Gene (41,0 kb)		
ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73		
Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Mikrozephalie: 27 Gene (88,3 kb)		
ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2		
Kabuki-Syndrom (KABUK) *, #	3 - 5 Wo	E
Gen-Panel: ID127.00, 2 Gene (20,8 kb)		
KDM6A, KMT2D		

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
Kleinwuchs, umfassende Diagnostik * Gen-Panel ID340.02 Kleinwuchs, umfassende Diagnostik: 207 Gene (511,0 kb) ACAN, ACP5, ACTB, ACTG1, AGPS, ALPL, AMMECR1, ANKRD11, ARCN1, ARSB, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BLM, BMP2, BMPR1B, BRAF, BRCA1, BRCA2, BRIP1, BTK, CBL, CCDC8, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CFAP410, COL10A1, COL11A2, COL27A1, COL2A, COMP, CREBBP, CRIPT, CSGALNACT1, CUL7, DDR2, DDRGK1, DHCR7, DNA2, DONSON, DPH1, EP300, ERCC4, ERCC6, ERCC8, EXOC6B, EXOSC2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGD1, FGFR1, FGFR3, FLNB, FN1, GALNS, GDF5, GH1, GHR, GHRHR, GHSR, GLB1, GLI2, GMNN, GNPAT, GPX4, GRHL2, GSC, GUSB, GZF1, HDAC8, HESX1, HMGA2, HRAS, HYAL1, IDUA, IGF1, IGF1R, IGF2, IGFALS, INSR, IRS1, IRS4, KIF22, KMT2A, KRAS, LARP7, LFNG, LHX3, LHX4, LTBP3, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MCM5, MRAS, NBAS, NBN, NEPRO, , NIPBL, NKX2-5, NKX3-2, NOTCH2, NPR2, NRAS, NSMCE2, OBSL1, ORC1, ORC4, ORC6, OTX2, PALB2, PAM16, PAPPA2, PAX8, PCNT, PEX5, PEX7, PHEX, PIK3R1, PISD, PLAG1, PLCB3, POC1A, POLR3GL, POP1, POU1F1, PPP1CB, PPP1R15B, PPP3CA, PRKG2, PRMT7, PROP1, PTH1R, PTPN11, PUS7, RAD21, RAD51, RAD51C, RAF1, RBBP8, RFWD3, RIT1, RMRP, RNPC3, RNU4ATAC, ROBO1, RPL13, RRAS2, RSPRY1, RTTN, SCUBE3, SGMS2, SHOC2, SHOX, SLC10A7, SLC26A2, SLX4, SMARCA2, SMC1A, SMC3, SOS1, SOS2, SOX, SOX3, SPRED2, SRCAP, STAT5B, TALDO1, TBCE, TBL1X, TBX15, TBX19, TBX2, THRA, TKT, TONSL, TOP3A, TRAIP, TRAPPC2, TRHR, TRIM37, TRIP11, TRMT10A , TRPV4, TSHB, TSHR, UBE2T, XRCC2, XRCC4 Wachstumshormonmangel (IGHD, CPHD): 14 Gene (23,0 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1, SOX3 Noonan-Syndrom (NS): 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Meier-Gorlin-Syndrom (MGORS): 8 Gene (12,6 kb) CDC45, CDC6, CDT1, GMNN, MCM5, ORC1, ORC4, ORC Seckel-Syndrom (SCKL): 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP Kongenitale Hypothyreose (CHNG): 8 Gene (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Skelettdysplasie (SED, SMD, AMD): 35 Gene (83,9 kb) ACAN, B3GALT6, BGN, BMPR1B, CFAP410, COL11A2, COL2A1, COMP, DDRGK1, DDR2, EXOC6B, FGFR3, FN1, GDF5, GPX4, KIF22, NEPRO, NKX3-2, NPR2, PAM16, PAPSS2, PISD, PLCB3, RMRP, RNU4ATAC, POP1, PRKG2, RPL13, RSPRY1, SIK3, SLC26A2, TONSL, TRAPPC2, TRIP11, TRPV4	4 - 6 Wo	E
Kongenitale Störung der Glykosylierung (CDG) * Gen-Panel ID035.02 Kongenitale Störung der Glykosylierung (CDG): 51 Gene (74,7 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3 Kongenitale Störung der Glykosylierung, Typ I (CDG1): 29 Gene (39,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Kongenitale Störung der Glykosylierung, Typ II (CDG2): 22 Gene (35,1 kb) ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199	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, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDFT1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNPK, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	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: ID147.00, 27 Gene (67,0 kb) ALDH18A1, ANAPC1, B3GALT6, B4GALT7, BANF1, BLM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LMNA, MDM2, MTX2, NAA10, POLD1, POLR3A, POLR3GL, PYCR1, RECQL4, SLC25A24, WRN, ZMPSTE24	3 - 5 Wo	E
RASopathien * Gen-Panel: ID015.05 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan-Syndrom (NS): 15 Gene (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Entwicklungs- und Wachstumsstörungen		
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
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
Sotos-Syndrom (SOTOS) und ähnliche Fehlbildungssyndrome * Gen-Panel: ID181.01, 4 Gene (18,8 kb) APC2, EZH2, NFIX, NSD1	3 - 5 Wo	E
Sprachentwicklungsstörung (DLD, CAS) * Gen-Panel: ID368.00 Sprachentwicklungsstörung (DLD, CAS): 34 Gene (119,7 kb) ATP2C2, BCL11A, BUD13, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, NFXL1, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TM4SF20, TNRC6B, UPF2, WDR5, ZFHX4, ZNF142, ZNF277 Kindliche Sprechapraxie (CAS): 29 Gene (110,2 kb) BCL11A, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TNRC6B, UPF2, WDR5, ZFHX4, ZNF142 Spezifische Sprachbeeinträchtigung (SLI): 5 Gene (9,5 kb) TP2C2, BUD13, NFXL1, TM4SF20, ZNF277	4 - 6 Wo	E
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
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
3M-Syndrom * Gen-Panel: ID214.00, 3 Gene (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 Wo	E
Epilepsien und Migräne		
Absence-Epilepsie (EJA, ECA) * Gen-Panel: ID057.02, 10 Gene (21,8 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, KCNMA1, RORB, SLC2A1, SLC12A5	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
Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE) * Gen-Panel ID080.03 Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), autosomal-dominant: 51 Gene (143,1 kb) ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CDK19, CELF2, CHD2, CUX2, CYFIP2, DNM1, EEF1A2, FBXO28, FGF12, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GNAO1, GRIN2B, GRIN2D, HCN1, HNRNPU, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, NEUROD2, NSF, NTRK2, PACS2, PHACTR1, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN2A, SCN3A, SCN8A, SIK1, SLC1A2, SPTAN1, STXBP1, YWHAG Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), autosomal-rezessiv: 45 Gene (105,0 kb) AARS1, ACTL6B, ADAM22, AP3B2, ARV1, CAD, CNPY3, CPLX1, DALRD3, DENND5A, DMXL2, DOCK7, FRRS1L, GAD1, GLS, GOT2, GRIN1, GUF1, HID1, ITPA, MDH1, MDH2, NECAP1, PARS2, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, SCN1B, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC38A3, ST3GAL3, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE), X-chromosomal: 9 Gene (20,3 kb) ALG13, ARHGEF9, ARX, CDKL5, FGF13, PCDH19 PIGA, SLC35A2, SMC1A	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Epilepsien und Migräne		
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
Epilepsien, umfassende Diagnostik * Gen-Panel ID061.05 Epilepsien, umfassende Diagnostik: 163 Gene (388,9 kb) AARS1, ACTL6B, ADAM22, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ASAH1, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, BRAT1, CACNA1A, CACNA1E, CACNB4, CAD, CASR, CDK19, CDKL5, CELF2, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CILK1, CLCN2, CLN8, CNPY3, CNTN2, CNTNAP2, CPA6, CPLX1, CSTB, CUX2, CYFIP2, DALRD3, DENND5A, DEPDC5, DMXL2, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GLS, GNAO1, GOT2, GOSR2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GUF1, HCN1, HCN2, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC1, KCNC2, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LGI1, LMNB2, MDH1, MDH2, MECP2, MEF2C, MTOR, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PLPBP, PPP3CA, PRDM8, PRICKLE1, PRRT2, RELN, RHOBTB2, RNF13, ROGDI, RORA, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC38A3, SLC6A1, SLC7A6OS, SMC1A, SNIP1, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Fokale Epilepsien: 17 Gene (50,9 kb) CHRNA2, CHRNA4, CHRNB2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24 Generalisierte Epilepsien: 39 Gene (72,6 kb) ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A6OS, SLC12A5, STX1B, TBC1D24 Epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP*, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG	4 - 6 Wo	E
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, ATP6V0A1, ATP6V1A, BRAT1, BSCL2, BTD, CACNA1A, CACNA1E, CAD, CCDC88A, CDK19, CDKL5, CELF2, CHD2, CLCN4, CNPY3, CPLX1, CNTNAP2, CUX2, CYFIP2, D2HGDH, DALRD3, DENND5A, DNM1, DNM1L, DMXL2, DOCK7, EEF1A2, ETHE1, FGF12, FGF13, FOLR1, FOXG1, FBXO28, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GAMT, GCSH, GLDC, GLS, GNAO1, GOT2, GPHN, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, IDH2, IFIH1, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, LIAS, MECP2, MEF2C, MDH1, MDH2, MFF, MOCS1, MOCS2, MTHFR, NAXD, NAXE, NECAP1, NEUROD2, NRXN1, NSF, NTRK2, PACS2, PARS2, PC, PCDH19, PHACTR1, PHGDH, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PPP3CA, PURA, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SERPINI1, SIK1, SLC1A2, SLC2A1, SLC12A5, SLC13A5, SLC6A8, SLC6A9, SLC9A6, SLC19A3, SLC25A1, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TCF4, TPK1, TRAK1, TREX1, UBA5, UGDH, UGP2, WDR45, WWOX, YWHAG Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Metabolische Enzephalopathie mit Epilepsie: 29 Gene (44,8 kb) ABAT, ADSL, ALDH5A1, ALDH7A1, AMT, BTD, D2HGDH, FOLR1, GAMT, GCSH, GLDC, GPHN, IDH2, LIAS, MDH2, MOCS1, MOCS2, MTHFR, PC, PHGDH, PNPO, POLG, SLC1A2, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, TPK1	4 - 6 Wo	E
Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFSP) * Gen-Panel: ID059.03 Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFSP): 9 Gene (35,9 kb) ADGRV1, CPA6, GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B Fieberkrämpfe (FEB): 5 Gene (30,3 kb) ADGRV1, CPA6, GABRG2, HCN2, SCN1A Generalisierte Epilepsie mit Fieberkrämpfen (GEFSP): 7 Gene (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 Wo	E
Fokale Epilepsien * Gen-Panel: ID208.01 Fokale Epilepsien: 17 Gene (50,9 kb) CHRNA2, CHRNA4, CHRNB2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24 Fokale Epilepsie mit variablen Herden (FFEVF): 4 Gene (13,7 kb) DEPDC5, NPRL2, NPRL3, SCN3A Nächtliche Frontallappenepilepsie (ENFL): 5 Gene (13,5 kb) CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNT1 Temporallappenepilepsie (ETL): 4 Gene (13,4 kb) CPA6, GAL, LGI1, RELN	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Epilepsien und Migräne		
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, SLC7A60S, SLC12A5, STX1B, TBC1D24 Myoklonusepilepsie (EJM, EPM): 24 Gene (45,3 kb) ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6B, SLC7A60S, TBC1D24 Absence-Epilepsie (EJA, ECA): 9 Gene (18,3 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, RORB, SLC2A1, SLC12A5 Generalisierte Epilepsie mit Fieberkrämpfen (GEFSP): 7 Gene (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 Wo	E
Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFSP, GEFS+) * Gen-Panel: ID235.02, 7 Gene (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 Wo	E
Hemiplegische Migräne (FHM) * Gen-Panel: ID064.01, 4 Gene (18,9 kb) ATP1A2, ATP1A3, CACNA1A, SCN1A	3 - 5 Wo	E
Metabolische Epilepsien * Gen-Panel: ID303.01 Metabolische Epilepsien: 84 Gene (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycin-Enzephalopathie (GCE): 5 Gene (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdän-Cofaktor-Defizienz (MOCOD): 3 Gene (3,7 kb) GPHN, MOCS1, MOCS2 Zerebrales Kreatinmangelsyndrom (CCDS): 3 Gene (3,9 kb) GAMT, GATM, SLC6A8 Ahornsirupkrankheit (MSUD): 5 Gene (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxy-Glutaraziduric: 3 Gene (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-Gangliosidose: 3 Gene (3,8 kb) HEXA, HEXB, GM2A Neuronale Ceroid-Lipofuszinose (CLN): 12 Gene (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 Wo	E
Migräne (MGR) * Gen-Panel: ID065.01 Migräne (MGR): 16 Gene (44,8 kb) ALPK1, ATP1A2, ATP1A3, CACNA1A, CSNK1D, ESR1, HTRA1, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1, TNF, TREX1 Familiäre hemiplegische Migräne (FHM): 3 Gene (16,6 kb) ATP1A2, CACNA1A, SCN1A	3 - 5 Wo	E
Myoklonusepilepsie (EPM, EJM) * Gen-Panel: ID116.03 Myoklonusepilepsie (EJM, EPM): 24 Gene (45,3 kb) ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6B, SLC7A60S, TBC1D24 Juvenile Myoklonusepilepsie (EJM): 12 Gene (28,0 kb) CACNB4, CASR, CILK1, CLCN2, EFHC1, GABRA1, GABRD, RORB, SCN1A, SLC2A1, SLC12A5, TBC1D24 Progressive Myoklonusepilepsie (EPM): 16 Gene (24,0 kb) ASAH1, CERS1, CLN8, CSTB, EPM2A, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SEMA6B, SLC7A60S	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Neugeborenenalter * Gen-Panel: ID135.00, 25 Gene (39,4 kb) ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter * Gen-Panel: ID171.00, 18 Gene (31,5 kb) ALDH5A1, ATP7A, BTD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1	3 - 5 Wo	E
Stoffwechselstörung mit Epilepsie im Schul- und Jugendalter * Gen-Panel: ID172.00, 15 Gene (28,1 kb) ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
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
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) * Gen-Panel: ID170.04, 37 Gene (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 Wo	E
Männliche Infertilität (SPGF, CBAVD) * Gen-Panel ID192.04 Männliche Infertilität (SPGF, CBAVD): 81 Gene (311,0 kb) ACTL9, ADGRG2, AK7, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC62, CEP112, CFAP43, CFAP44, CFAP47, CFAP58, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CFTR, DNAH1, DNAH2, DNAH8, DNAH10, DNAH17, DNHD1, DPY19L2, DZIP1, FANCM, FBXO43, FSIP2, IFT74, GCNA, GGN, KLHL10, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPAG17, SPATA16, SPEF2, SPINK2, STAG3, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TERB1, TERB2, TEX11, TEX14, TEX15, TDRD9, TSGA10, TTC21A, TTC29, USP9Y, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Störung der Spermatogenese (SPGF): 79 Gene (303,5 kb) ACTL9, AK7, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC62, CEP112, CFAP43, CFAP44, CFAP47, CFAP58, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, DNAH1, DNAH2, DNAH8, DNAH10 DNAH17, DNHD1, DPY19L2, DZIP1, FANCM, FBXO43, FSIP2, IFT74, GCNA, GGN, KLHL10, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPAG17, SPATA16, SPEF2, SPINK2, STAG3, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TERB1, TERB2, TEX11, TEX14, TEX15, TDRD9, TSGA10, TTC21A, TTC29, USP9Y, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Kongenitale bilaterale Aplasie des Vas deferens (CBAVD): 2 Gene (7,5 kb) ADGRG2, CFTR	4 - 6 Wo	E
Ovarialdysgenesie (ODG) * Gen-Panel: ID293.01 Ovarialdysgenesie (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX-Gonadendysgenesie (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX-Gonadendysgenesie mit Taubheit (PRLTS): 6 Gene (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 Wo	E
Störung der Geschlechtsentwicklung (DSD) * Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NR0B1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSPO1, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NR0B1, POR, PPP1R12A, PPP2R3C, RSPO1, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
Vorzeitige Ovarialinsuffizienz (POF) * Gen-Panel: ID078.05 Vorzeitige Ovarialinsuffizienz (POF): 23 Gene (51,1 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, INHA, LHCGR, MCM8, MSH5, NOBOX, NR5A1, POF1B, STAG3, SYCE1, XRCC2 Vorzeitige Ovarialinsuffizienz (POF), X-chromosomal: 4 Gene (8,2 kb) BMP15, DIAPH2, FMR1, POF1B Vorzeitige Ovarialinsuffizienz (POF), autosomal-rezessiv: 10 Gene (25,0 kb) C14ORF39, GDF9, FANCM, HFM1*, HSF2BP, MCM8*, MSH5, STAG3, SYCE1, XRCC2 Vorzeitige Ovarialinsuffizienz (POF), autosomal-dominant: 6 Gene (12,7 kb) BNC1, ERCC6, FIGLA, FOXL2, NOBOX, NR5A1	3 - 5 Wo	E
Weibliche Infertilität (PREMBL, OOMD) * Gen-Panel: ID239.01, 12 Gene (18,0 kb) BTG4, PADI6, PANX1, PATL2, REC114, TLE6, TRIP13, TUBB8, WEE2, ZP1, ZP2, ZP3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Fettstoffwechselstörungen		
Fettstoffwechselstörungen, umfassende Diagnostik * Gen-Panel: ID044.02, 56 Gene (107,1 kb) ABCA1, ABCG5, ABCG8, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA1, APOA2, APOA4, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CETP, CIDEC, CREB3L3, CYP27A1, CYP7A1, DHCR7, DHCR24, GCKR, GK, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIMA1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPA, LPL, MTTP, NPC1, NPC1L1, NPC2, PCSK9, PLIN1, PNPLA5, POLD1, PPARG, SAR1B, SCARB1, SMPD1, SORT1, ZMPSTE24	4 - 6 Wo	E
Fettstoffwechselstörung durch LDL-Mangel * Gen-Panel: ID094.01, 10 Gene (29,6 kb) ANGPTL3, ANGPTL4, ANGPTL8, APOB, APOE, LIMA1, MTTP, NPC1L1, PCSK9, SAR1B	3 - 5 Wo	E
HDL-assoziierte Fettstoffwechselstörung * Gen-Panel: ID096.01, 15 Gene (25,5 kb) ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA2, APOA4, APOC3, CETP, LCAT, LIPC, LIPG, NPC1, NPC2, SCARB1, SMPD1	3 - 5 Wo	E
Hypercholesterinämie * Gen-Panel: ID024.02, 13 Gene (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1	3 - 5 Wo	E
Hypertriglyzeridämie * Gen-Panel:ID095.02 Hypertriglyzeridämie: 29 Gene (51,1 kb) AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEC, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PLIN1, POLD1, PPARG, ZMPSTE24 Hyperchylomikronämie: 6 Gene (6,1 kb) APOC2, APOE, APOA5, GPIHBP1, LMF1, LPL Lipodystrophie: 12 Gene (19,0 kb) AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, LIPE, LMNA, PLIN1, POLD1, PPARG, ZMPSTE2	3 - 5 Wo	E
Kombinierte Hyperlipidämie * Gen-Panel: ID025.06 Kombinierte Hyperlipidämie: 40 Gene (72,8 kb): ABCG5, ABCG8, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEC, CREB3L3, CYP27A1, CYP7A1, GCKR, GK, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, NPC1L1, PCSK9, PLIN1, PNPLA5, POLD1, PPARG, SORT1, ZMPSTE24 Hypercholesterinämie: 13 Gene (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP7A1, CYP27A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1 Hypertriglyzeridämie: 29 Gene (51,1 kb) AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEC, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PLIN1, POLD1, PPARG, ZMPSTE24	3 - 5 Wo	E
Lipodystrophie, umfassende Diagnostik * Gen-Panel: ID343.00, 45 Gene (80,0 kb) ADRA2A, AGPAT2, AKT2, ALDH18A1, BANF1, BSCL2, CAV1, CAVIN1, CIDEC, EPHX1, ERCC6, ERCC8, FBN1, HRAS, KCNJ6, LEP, LEPR, LIPE, LMNA, MCM3, MCM7, MFN2, MTX2, NSMCE2, OPA3, OTULIN, PCYT1A, PDGFRB, PIK3R1, PLIN1, POLD1, POLR3A, POMP, PPARG, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PTPN11, SLC25A24, SLC29A3, SPRTN, WRN, ZMPSTE24	3 - 5 Wo	E
Lipodystrophie * Gen-Panel: ID055.03 Lipodystrophie (CGL, FPLD): 12 Gene (19,0 kb) AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, LIPE, LMNA, PLIN1, POLD1, PPARG, ZMPSTE24 Kongenitale generalisierte Lipodystrophie (CGL): 4 Gene (3,7 kb) AGPAT2, BSCL2, CAV1, CAVIN1 Familiäre partielle Lipodystrophie (FPLD): 6 Gene (9,6 kb) CAV1, CIDEC, LIPE, LMNA, PLIN1, PPARG	3 - 5 Wo	E
Statin-assoziierte Myopathie * Gen-Panel: ID097.00, 10 Gene (35,2 kb) ACADM, ACADS, ACADVL, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1	3 - 5 Wo	E
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
Glykogenspeicherkrankheit (GSD) * Gen-Panel: ID108.00, 23 Gene (46,9 kb) AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Gastroenterologische Erkrankungen		
Intrahepatische Cholestase * Gen-Panel: ID159.04 Intrahepatische Cholestase: 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 ID006.08 Kolorektales Karzinom und Polyposis: 22 Gene (62,3 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Kolorektale Polyposis: 14 Gene (37,6 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11 Kolorektales Karzinom: 14 Gene (40,4 kb) ATM, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11	3 - 5 Wo	E
Lynch-Syndrom (LYNCH, HNPCC) * Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	2 - 4 Wo	E
Magenkarzinom * Gen-Panel: ID090.03, 24 Gene (71,9 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, 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: ID089.03, 17 Gene (55,2 kb) APC, ATM, BRCA1, BRCA2, CDKN2A, CTRC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL	3 - 5 Wo	E
Pankreatitis (PCTT) * Gen-Panel: ID141.02, 18 Gene (29,6 kb) APOA5, APOC2, CASR, CEL, CFTR, CPA1, CLDN2, CELA3B, CTRC, CTSB, GPIHBP1, LMF1, LPL, PNLIP, PRSS1, SPINK1, TRPV6, UBR1	3 - 5 Wo	E
Polyposis-Syndrom (PS, FAP) * Gen-Panel: ID005.05, 14 Gene (37,6 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
Polyzystische Lebererkrankung (PCLD) * Gen-Panel: ID305.01 Polyzystische Lebererkrankung (PCLD): 11 Gene (45,6 kb) ALG5, ALG8, DNAJB11, DZIP1L, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63 Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 4 Gene (10,3 kb) ALG8, LRP5, PRKCSH, SEC63 Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 7 Gene (35,3 kb) ALG5, DNAJB11, DZYP1L, GANAB, PKD1, PKD2, PKHD1	3 - 5 Wo	E
Viszerale Myopathien und Neuropathien, umfassende Diagnostik * Gen-Panel: ID238.02 Viszerale Myopathien und Neuropathien, umfassende Diagnostik: 36 Gene (83,7 kb) ACTA2, ACTG2, CHRM3, DNMT3B, ECE1, EDN3, EDNRB, ERBB2, ERBB3, FLNA, GDNF, GFRA1, KIFBP, L1CAM, LIG3, LMOD1, MYH11, MYL9, MYLK, NRG1, NRTN, NTF3, NTRK3, PHOX2B, POLG, PSPN, RAD21, RET, RRM2B, SEMA3A, SEMA3C, SEMA3D, SGO1, SOX10, TYMP, ZEB2 Morbus Hirschsprung (HSCR): 20 Gene (37,1 kb) ECE1, EDN3, EDNRB, DNMT3B, GDNF, GFRA1, KIFBP, L1CAM, NRG1, NRTN, NTF3, NTRK3, PHOX2B, PSPN, RET, SEMA3A, SEMA3C, SEMA3D, SOX10, ZEB2 Neuropathische intestinale Pseudoobstruktion (VSCN): 9 Gene (28,6 kb) ERBB2, ERBB3, FLNA, LIG3, POLG, RAD21, RRM2B, SGO1, TYMP Myopathische intestinale Pseudoobstruktion (VSCM): 8 Gene (19,9 kb) ACTA2, ACTG2, CHRM3, LMOD1, MYL9, MYLK, MYH11, RAD21	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Gehirnfehlbildungen		
Aicardi-Goutières-Syndrom (AGS) * Gen-Panel: ID058.01, 9 Gene (13,0 kb) 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, TOGAGRAM1, TTC21B, ZNF423	4 - 6 Wo	E
Komplexe kortikale Dysplasie mit weiteren Gehirnfehlbildungen (CDCBM) * Gen-Panel: ID271.01, 13 Gene (47,1 kb) APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1	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, FKRP, FKTN, FLNA, GMPPB, KATNB1, KIFBP, KIF2A, KIF5C, KIF26A, LAMB1, LAMC3, LARGE1, MACF1, MAP1B, MTOR, NDE1, NEDD4L, OCLN, PAFAH1B1, PHGDH, PI4KA, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSAT1, RAB18, RAB3GAP1, RAB3GAP2, RAC3, RELN, RTTN, RXYLT1, SHH, SIX3, TBC1D20, TMTC3, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2, WDR62 Periventrikuläre noduläre Heterotopie (PVNH): 6 Gene (26,1 kb) ARF1, ARFGEF2, ERMARD, FLNA, MAP1B, NEDD4L Komplexe kortikale Dysplasie (CDCBM): 12 Gene (45,7 kb) APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1 Walker-Warburg-Syndrom (MDDGA): 14 Gene (23,7 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Lissenzephalie (LIS): 12 Gene (46,4 kb) ARX, CDK5, CEP85L, DCX, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A Polymikrogyrie: 15 Gene (42,0 kb) ADGRG1, AKT3, CCND2, COL3A1, FIG4, KIFBP, OCLN, PI4KA, PIK3CA, RTTN, PIK3R2, TUBA1A, TUBA8, TUBB2B, WDR62 Schizenzephalie: 7 Gene (19,7 kb) COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62	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
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Zerebelläre Dysgenesie, 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		
Atypisches hämolytisch-urämisches Syndrom (AHUS) *	3 - 5 Wo	E
Gen-Panel: ID163.04, 20 Gene (42,1 kb)		
ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN		
Erythrozytose (ECYT) *	3 - 5 Wo	E
Gen-Panel: ID138.02, 11 Gene (13,9 kb)		
BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL		
Fanconi-Anämie (FANC) *	3 - 5 Wo	E
Gen-Panel: ID043.02, 21 Gene (60,7 kb)		
BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2		
Hämochromatose (HFE) und Hämosiderose *	3 - 5 Wo	E
Gen-Panel: ID114.04, 10 Gene (14,6 kb)		
BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2		
Hämophilie *	3 - 5 Wo	E
Gen-Panel: ID154.00, 3 Gene (16,9 kb)		
F8, F9, VWF		
Hermansky-Pudlak-Syndrom (HPS) *	3 - 5 Wo	E
Gen-Panel: ID289.00, 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Knochenmarkinsuffizienz (BMF) *	3 - 5 Wo	E
Gen-Panel: ID357.00		
Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb):		
ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTEL1, SRP72, TERC, TERT, TP53, ZCCHC8		
Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb)		
ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53		
Telomer-assozierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb)		
PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) *	4 - 6 Wo	E
Gen-Panel: ID321.01		
Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML): 121 Gene (244,1 kb):		
ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8		
Akute myeloische Leukämie (AML): 12 Gene (28,4 kb)		
ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53		
Diamond-Blackfan-Anämie (DBA): 20 Gene (11,3 kb)		
GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2		
Shwachman-Diamond-Syndrom (SDS): 4 Gene (7,2 kb)		
DNAJC21, EFL1, SBDS, SRP54		
Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb)		
ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53		
Lungenfibrose und Knochenmarkinsuffizienz (PFBMFT): 6 Gene (13,0 kb)		
PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8		
Dyskeratosis congenita (DKC): 13 Gene (21,8 kb)		
ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53		
Kongenitale Neutropenie (SCN): 10 Gene (13,9 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS		
Fanconi-Anämie (FANC): 20 Gene (60,7 kb)		
BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2		
Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb)		
MLH1, MSH2, MSH6, PMS2		
Neutropenie *	3 - 5 Wo	E
Gen-Panel: ID189.02		
Neutropenie: 30 Gene (51,0 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1		
Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb)		
CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS		
Syndromale Erkrankung mit Neutropenie: 21 Gene (37,5 kb)		
ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, USB1, VPS13B, WDR1		
Porphyrie *	3 - 5 Wo	E
Gen-Panel: ID153.01, 10 Gene (12,8 kb)		
ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS		

Erkrankung/Diagnostik	Dauer	Material
Hämatologische Erkrankungen		
Sideroblastische Anämie (SIDBA) *	3 - 5 Wo	E
Gen-Panel: ID355.00, 10 Gene (14,9 kb)		
ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2		
Sphärozytose (SPH) und Elliptozytose (EL) *	3 - 5 Wo	E
Gen-Panel: ID203.01		
Sphärozytose (SPH) und Elliptozytose (EL): 6 Gene (26,8 kb)		
ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB		
Sphärozytose (SPH): 5 Gene (24,2 kb)		
ANK1, EPB42, SLC4A1, SPTA1, SPTB		
Elliptozytose (EL): 4 Gene (18,4 kb)		
EPB41, SLC4A1, SPTA1, SPTB		
Thrombophilie (THPH) *	3 - 5 Wo	E
Gen-Panel: ID150.01, 12 Gene (25,2 kb)		
F2, F5, F9, F13B, HABP2, HRG, MTHFR, PROC, PROS1, SERPINC1, SERPIND1, THBD		
Thrombotische Mikroangiopathie (TMA) *	3 - 5 Wo	E
Gen-Panel: ID707.00		
Thrombotische Mikroangiopathie (TMA): 23 Gene (44,7 kb)		
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD		
Thrombotisch-thrombozytopenische Purpura (TTP): 1 Gen (4,3 kb)		
ADAMST13		
Atypisches hämolytisch-urämisches Syndrom (AHUS): 23 Gene (44,7 kb)		
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PLG, THBD		
Thrombozytendefekte, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID274.01		
Thrombozytendefekte, umfassende Diagnostik: 64 Gene (142,3 kb)		
ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, EPHB2, ETV6, FERMT3, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB3, JAK2, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TUBB1, WAS, WDR1, WIPF1		
Thrombozytopenie, nicht-syndromal (THC): 9 Gene (17,7 kb)		
ANKRD26, CYCS, ETV6, FYB1, IKZF5, MASTL, SRC, TUBB1, WAS		
Thrombozythämie (THCYT): 3 Gene (6,4 kb)		
JAK2, MPL, THPO		
Thrombozytenbedingte Blutungsstörung (BDPLT): 23 Gene (52,8 kb)		
ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1		
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Thrombozytopathie (BDPLT, HPS) *	3 - 5 Wo	E
Gen-Panel: ID119.01		
Thrombozytopathie (BDPLT, HPS): 37 Gene (91,0 kb)		
ACTN1, ANO6, AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CD36, DTNBP1, EPHB2, FERMT3, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1		
Thrombozytenbedingte Blutungsstörung (BDPLT) mit Thrombozytopenie: 15 Gene (37,9 kb)		
ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14		
Thrombozytenbedingte Blutungsstörung (BDPLT) ohne Thrombozytopenie: 10 Gene (20,3 kb)		
ANO6, EPHB2, GP6, ITGA2B, ITGB3, P2RY12, PTGS1, RASGRP2, TBXA2R, TBXAS1		
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Thrombozytopenie *	3 - 5 Wo	E
Gen-Panel: ID104.02		
Thrombozytopenie (THC): 40 Gene (84,7 kb)		
ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CD36, CDC42, CYCS, DIAPH1, ETV6, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP9, HOXA11, IKZF5, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, PLAU, PRKACG, RBM8A, RUNX1, SLFN14, SRC, STIM1, TUBB1, WAS, WDR1, WIPF1		
Thrombozytopenie, nicht-syndromal (THC): 9 Gene (17,7 kb)		
ANKRD26, CYCS, ETV6, FYB1, IKZF5, MASTL, SRC, TUBB1, WAS		
Thrombozytenbedingte Blutungsstörung (BDPLT): 15 Gene (37,9 kb)		
ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14		

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Adams-Oliver-Syndrom (AOS) *		
Gen-Panel: ID259.00, 6 Gene (23,2 kb)	3 - 5 Wo	E
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		
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	3 - 5 Wo	E
Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Waardenburg-Syndrom (WS): 8 Gene (9,3 kb)		
EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR		
Griscelli-Syndrom (GS): 3 Gene (8,0 kb)		
MLPH, MYO5A, RAB27A		
Amelogenesis imperfecta (AI) *		
Gen-Panel: ID232.01, 17 Gene (29,4 kb)	3 - 5 Wo	E
ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAPH, RELT, SLC24A4, WDR72		
Cowden-Syndrom (CWS) *		
Gen-Panel: ID075.01, 8 Gene (12,8 kb)	3 - 5 Wo	E
AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1		
Cutis laxa (ARCL, ADCL) *		
Gen-Panel: ID109.02, 11 Gene (27,9 kb)	3 - 5 Wo	E
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
Dyskeratosis congenita (DKC) *		
Gen-Panel: ID347.01, 15 Gene (24,1 kb)	3 - 5 Wo	E
ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53		
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	3 - 5 Wo	E
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), nicht-syndromal: 20 Gene (27,5 kb)		
CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A		
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), syndromal: 28 Gene (51,0 kb)		
AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2		
Ektodermale Dysplasie (ECTD), umfassende Diagnostik *		
Gen-Panel: ID366.00		
Ektodermale Dysplasie (ECTD), 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)	4 - 6 Wo	E
CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A		
Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), syndromal: 28 Gene (51,0 kb)		
AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2		
Komplexe Syndrome mit ektodermaler Dysplasie: 42 Gene (120,5 kb)		
ANTXR1, ARID1A, ARID1B, ATP6V1B2, CDH1, CTNND1, CTSK, DSG4, DSP, EVC, EVC2, FGF10, FGFR2, FGFR3, HEPHL1, IFT122, IFT140, IFT43, IFT52, INSR, KCTD1, KRT14, KRT16, KRT17, KRT81, KRT83, KRT86, NLRP1, PEX1, PEX6, ROGDI, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SREBF1, TBC1D24, UBR1, WDR19, WDR35		
Epidermolysis bullosa (EB) *		
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)	4 - 6 Wo	E
COL17A1, ITGA3, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2		
Epidermolysis bullosa dystrophica (DEB): 1 Gen (8,8 kb)		
COL7A1		
Kindler Epidermolysis bullosa (KNDLRS): 1 Gen (2,0 kb)		
FERMT1		
Syndromale Epidermolysis bullosa: 20 Gene (63,9 kb)		
ATP2A2, ATP2C1, CAST, CD151, CDSN, CHST8, CSTA, DSG1, DSP, FERMT1, FLG2, IKBKG, ITGA3, JUP, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5		

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Gorlin-Goltz-Syndrom (BCNS) *	2 - 4 Wo	E
Gen-Panel: ID174.00, 3 Gene (9,4 kb)		
PTCH1, PTCH2, SUFU		
Hereditäres Angioödem (HAE) *	3 - 5 Wo	E
Gen-Panel: ID345.00, 7 Gene (16,4 kb)		
ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1		
Hermansky-Pudlak-Syndrom (HPS) *	3 - 5 Wo	E
Gen-Panel: ID289.00, 11 Gene (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Hydrops fetalis *	3 - 5 w	E
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		
Hypotrichose, nicht-syndromale Form (HYPT) *	3 - 5 Wo	E
Gen-Panel: ID146.01, 12 Gene (20,1 kb)		
APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE		
Ichthyose *	3 - 5 Wo	E
Gen-Panel: ID217.01		
Ichthyose: 38 Gene (73,0 kb)		
ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CLDN10, CSTA, CYP4F22, ELOVL4, ERCC2, FLG, GJB2, KRT1, KRT10, KRT2, LIPN, LORICRIN, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, TGM1		
Kongenitale Ichthyose, autosomal-rezessiv (ARCI): 14 Gene (28,7 kb)		
ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1		
Ichthyose, autosomal-dominant und X-chromosomal: 7 Gene (21,3 kb)		
ASPRV1, FLG, GJB2, KRT1, KRT2, KRT10, STS		
Kutanes malignes Melanom (CMM) *	3 - 5 Wo	E
Gen-Panel: ID193.01, 12 Gene (26,4 kb)		
BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3		
Multiples Pterygium-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID158.01, 8 Gene (16,8 kb)		
CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4		
Neurofibromatose (NF) *, #	3 - 5 Wo	E
Gen-Panel: ID210.00, 3 Gene (11,6 kb)		
NF1, NF2, SPRED1		
Okulokutaner Albinismus (OCA) *	3 - 5 Wo	E
Gen-Panel: ID082.02, 9 Gene (13,3 kb)		
DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Orofaziodigitales Syndrom (OFD) *	3 - 5 Wo	E
Gen-Panel: ID265.00, 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Pachyonychia congenita (PC) *	3 - 5 Wo	E
Gen-Panel: ID120.01, 13 Gene (30,1 kb)		
AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3		
Palmoplantarkeratose (PPK) *	3 - 5 Wo	E
Gen-Panel: ID323.00		
Palmoplantarkeratose (PPK): 33 Gene (58,2 kb)		
AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB3, GJB4, JUP, KDSR, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT10, KRT14, KRT16, KRT17, KRT83, LORICRIN, MBTPS2, PERP, RHBDF2, SERPINB7, SLURP1, SMARCAD1, TAT, TRPM4, TRPV3, WNT10A		
Palmoplantarkeratose (PPK), nicht-syndromal: 11 Gene (25,7 kb)		
AAGAB, AQP5, DSG1, DSP, KRT1, KRT6C, KRT9, KRT10, KRT16, SERPINB7, TRPV3		
Palmoplantarkeratose (PPK), syndromal: 15 Gene (22,4 kb)		
CTSC, ENPP1, GJA1, GJB2, KRT14, MBTPS2, JUP, LORICRIN, PERP, RHBDF2, SLURP1, SMARCAD1, TAT, TRPV3, WNT10A		
Erythrokeratodermia variabilis et progressiva (EKVP): 7 Gene (9,5 kb)		
GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4		
Pachyonychia congenita (PC): 4 Gene (6,1 kb)		
KRT6A, KRT6B, KRT16, KRT17		
Peeling-Skin-Syndrom (PSS) *	3 - 5 Wo	E
Gen-Panel: ID309.00, 6 Gene (13,6 kb)		
CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5		
Pierre-Robin-Sequenz *	3 - 5 Wo	E
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		

Erkrankung/Diagnostik	Dauer	Material
Haut- und Zahnerkrankungen		
Progerie und progeroide Syndrome * Gen-Panel: ID147.00, 27 Gene (67,0 kb) ALDH18A1, ANAPC1, B3GALT6, B4GALT7, BANF1, BLM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LMNA, MDM2, MTX2, NAA10, POLD1, POLR3A, POLR3GL, PYCR1, RECQL4, SLC25A24, 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: ID010.02, 15 Gene (143,9 kb) DH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, PLN, LMNA, PKP2, PRKAG2, TGFB3, TMEM43, TTN	4 - 6 Wo	E
Atriumseptumdefekt und Ventrikelseptumdefekt (ASD, VSD, AVSD) * Gen-Panel: ID143.00, 12 Gene (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1	3 - 5 Wo	E
Bikuspide Aortenklappe (AOVD) * Gen-Panel: ID301.00, 6 Gene (16,7 kb) GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2	3 - 5 Wo	E
Bindegewebserkrankungen mit Aortenbeteiligung * Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, TNXB, ZNF469 Thorakales Aortenaneurysma und Aortendissektion (TAA/D): 17 Gene (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFBR2, TGFBR1 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
Brugada-Syndrom (BRGDA) * Gen-Panel: ID014.02, 23 Gene (56,4 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4	3 - 5 Wo	E
CADASIL und CARASIL *, # Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E
CHARGE-Syndrom * Gen-Panel: ID307.00, 3 Gene (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 Wo	E
Cutis laxa (ARCL, ADCL) * Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Dilatative Kardiomyopathie (CMD, DCM) *		
Gen-Panel: ID008.03		
Dilatative Kardiomyopathie (CMD, DCM): 63 Gene (287,5 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CRYAB, CSRP3, DES, DMD, DNAJC19, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GATAD1, GET3, HFE, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, SYNE2, TAFAZZIN, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TBX20, TTN, VCL, VEZF1		
Dilatative Kardiomyopathie, nicht-syndromal, dominant (CMD1): 42 Gene (201,2 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DSG2, EYA4, FKTN, FLNC, LAMA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PLN, PRDM16, PSEN1, PSEN2, RAF1, RBM20, SCN5A, SDHA, SGCD, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TBX20, TTN, VCL, VEZF1	4 - 6 Wo	E
Dilatative Kardiomyopathie, nicht-syndromal, rezessiv (CMD2): 8 Gene (9,7 kb)		
BAG5, GATAD1, GET3, JPH2, LMOD2, PPCS, RPL3L, TNNI3		
Dilatative Kardiomyopathie, nicht-syndromal, X-chromosomal (CMD3): 2 Gene (11,9 kb)		
DMD, TAFAZZIN		
Dilatative Kardiomyopathie, syndromal: 18 Gene (86,9 kb)		
DES, DMD, DNAJC19, DSP, EMD, FHL1, FKRP, HFE, JUP, LAMP2, LMNA, MYH7, MYL2, SYNE1, SYNE2, TAFAZZIN, TCAP, TMEM43		
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	3 - 5 Wo	E
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		
Endokrine Hypertonie *		
Gen-Panel: ID270.02		
Endokrine Hypertonie: 32 Gene (80,9 kb)		
CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, WNK1, WNK4, YY1AP1		
Hyperaldosteronismus (HALD): 6 Gene (20,6 kb)		
CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5		
Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb)	3 - 5 Wo	E
CUL3, KLHL3, WNK1, WNK4		
Phäochromozytom/Paragangliom-Syndrom (PPGL): 9 Gene (9,5 kb)		
MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, 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		
Fallot-Tetralogie (TOF) *		
Gen-Panel: ID144.01, 12 Gene (23,5 kb)	3 - 5 Wo	E
FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2		
Familiäres Vorhofflimmern (ATFB) *		
Gen-Panel: ID016.02, 17 Gene (29,5 kb)	3 - 5 Wo	E
ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A		
Frühes Repolarisationssyndrom (ERS) *		
Gen-Panel: ID330.00, 12 Gene (38,9 kb)	3 - 5 Wo	E
ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A		
Hereditäre hämorrhagische Teleangiektasie (HHT) *		
Gen-Panel: ID155.01, 7 Gene (15,6 kb)	3 - 5 Wo	E
ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4		
Herz-Hand-Syndrom *		
Gen-Panel: ID165.01, 9 Gene (23,8 kb)	3 - 5 Wo	E
DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5		
Hypertrophe Kardiomyopathie (CMH, HCM) *		
Gen-Panel: ID007.03		
Hypertrophe Kardiomyopathie (HCM, CMH): 56 Gene (209,1 kb)		
ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CAV3, CSRP3, DES, FHL1, FHOD3, FLNC, GAA, GLA, HRAS, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RRAS2, RIT1, SOS1, SOS2, SPRED2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL		
Hypertrophe Kardiomyopathie, nicht-syndromal (CMH): 35 Gene (177,2 kb)	4 - 6 Wo	E
ACTC1, ACTN2, ALPK3, ANKRD1, CAV3, CSRP3, DES, FHL1, FHOD3, FLNC, JPH2, KLF10, KLHL24, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, VCL		
Hypertrophe Kardiomyopathie, syndromal: 22 Gene (33,6 kb)		
BRAF, CALR3, GAA, GLA, HRAS, KRAS, LAMP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NRAS, PRKAG2, PTPN11, RAF1, RRAS2, RIT1, SOS1, SOS2, SPRED2, TTR		
Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) *		
Gen-Panel: ID012.03, 9 Gene (33,9 kb)	3 - 5 Wo	E
ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN		

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Kardiale Arrhythmien, umfassende Diagnostik * Gen-Panel: ID026.03 Kardiale Arrhythmien, umfassende Diagnostik: 71 Gene (274,2 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, EMD, FGF12, GJA5, GNB2, GNB5, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYH6, MYL4, NKX2-5, NPPA, NUP155, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TANGO2, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN Long-QT-Syndrom (LQT): 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN Brugada-Syndrom (BRGDA): 23 Gene (56,4 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPDL, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2V, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4 Frühes Repolarisationssyndrom (ERS): 12 Gene (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A Familiäres Vorhofflimmern (ATFB): 17 Gene (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A Short-QT-Syndrom (SQT): 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SCL4A3 Sick-Sinus-Syndrom (SSS): 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT): 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN Arrhythmogene rechtsventrikuläre Dysplasie (ARVD): 15 Gene (150,6 kb) CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, PLN, LMNA, PKP2, PRKAG2, RYR2, TGFB3, TMEM43, TTN	4 - 6 Wo	E
Kardiomyopathien, umfassende Diagnostik * Gen-Panel: ID027.02 Kardiomyopathien, umfassende Diagnostik: 132 Gene (456,7 kb) ABCC9, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, BAG3, BAG5, BRAF, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, COA5, COA6, COX15, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FXN, GAA, GATAD1, GET3, GLA, HADHA, HCN4, HFE, HRAS, ILK, JPH2, JUP, KIF20A, KLF10, KLHL24, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LIMS2, LMNA, LMOD2, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM10, MIB1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RBM20, RIT1, RPL3L, RRAS2, RYR2, SCN2B, SCN5A, SCO2, SDHA, SGCB, SGCD, SGCG, SHOC2, SLC25A4, SOS1, SOS2, SPRED2, SYNE1, SYNE2, TAFAZZIN, TBX20, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, VEZF1 Dilatative Kardiomyopathie (DCM, CMD): 63 Gene (287,5 kb) ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CRYAB, CSRP3, DES, DMD, DMAJC19, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GATAD1, GET3, HFE, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, SYNE2, TAFAZZIN, TBX20, TCP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1 Hypertrophe Kardiomyopathie (HCM, CMH): 56 Gene (209,1 kb) ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CAV3, CSRP3, DES, FHL1, FHOD3, FLNC, GAA, GLA, HRAS, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RRAS2, SOS1, SOS2, SPRED2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL Restriktive Kardiomyopathie (RCM): 14 Gene (31,4 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MCM10, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC): 15 Gene (150,6 kb) CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, LMNA, PKP2, PLN, PRKAG2, RYR2, TGFB3, TMEM43, TTN Linksventrikuläre Noncompaction-Kardiomyopathie (LVNC, NCCM): 18 Gene (169,0 kb) ACTC1, ACTN2, DMD, DTNA, HCN4, LDB3, LMNA	4 - 6 Wo	E
Kongenitale Herzfehler, umfassende Diagnostik * Gen-Panel: ID019.02 Kongenitale Herzfehler, umfassende Diagnostik: 149 Gene (472,9 kb) ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR2B, ADAMTS10, ADAMTS17, ADAMTS19, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CFAP45, CFAP52, CFAP53, CFC1, CHD4, CHD7, CIROP, CITED2, CREBBP, CRELD1, DHCR7, DLL4, DNAAF1, DNAH5, DNAH9, DNAH11, DOCK6, DPYSL5, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FLT4, FOXC1, FOXF1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAAO, HAND1, HOXA1, HRAS, ISL1, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MMP21, MNS1, MRAS, MYH11, MYH6, MYRF, NADSYN1, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2, NR2F2, NRAS, NSD1, ODAD2, PIGL, PITX2, PKD1L1, PLD1, PPP1CB, PRDM6, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, ROBO4, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMAD6, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, SPRED2, STAG2, STRA6, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TKT, TLL1, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZFPM2, ZIC3 Isolierte kongenitale Herzfehler: 48 Gene (149,9 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Syndromale kongenitale Herzfehler: 109 Gene (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Kongenitale isolierte Herzfehler * Gen-Panel: ID017.04 Kongenitale isolierte Herzfehler: 43 Gene (103,7 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Atrium-/Ventrikelseptumdefekt (ASD, VSD): 12 Gene (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NR2F2, NKX2-5, TBX20, TLL1 Fallot-Tetralogie (TOF): 12 Gene (23,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2 Viszerale Heterotaxie (HTX): 13 Gene (26,2 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, MMP21, MNS1, NODAL, PKD1L1, ZIC3 Konotrunkale Herzfehlbildung (CTHM): 12 Gene 24,8 (kb) FLT4, FOXH1, CFC1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3 Aortenklappenerkrankung (AOVD): 7 Gene (19,1 kb) ELN, GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2 Hypoplastisches Linksherzsyndrom (HLHS): 5 Gene (5,2 kb) GDF1, GJA1, HAND1, NKX2-5, NR2F2 Kongenitale multiple Herzfehlbildung (CHTD): 7 Gene (14,4 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, TAB2, ZIC3 Herzklappendysplasie (CVDP): 3 Gene (14,8 kb) ADAMTS19, FLNA, PLD1	4 - 6 Wo	E
Kongenitale syndromale Herzfehler * Gen-Panel ID252.02 Kongenitale synromale Herzfehler: 109 Gene (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFBR1, TGFBR2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3 Noonan-Syndrom (NS): 16 Gene (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 Ritscher-Schinzel-Syndrom (RTSC): 4 Gene (10,0 kb) DPYSL5, CCDC22, VPS35L, WASHC5 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Adams-Oliver-Syndrom (AOS): 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ Kabuki-Syndrom (KABUK): 2 Gene (20,8 kb) KDM6A, KMT2D Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 CHARGE-Syndrom: 2 Gene (11,3 kb) CHD7, SEMA3E VCRL-Syndrom: 3 Gene (4,4 kb) HAAO, KYNU, NADSYN1	4 - 6 Wo	E
Kongenitale multiple Herzfehlbildung (CHTD) * Gen-Panel: ID302.00, 7 Gene (14,4 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, TAB2, ZIC3	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
Linksventrikuläre Noncompaction-Kardiomyopathie (LVNC, NCCM) * Gen-Panel: ID011.02, 18 Gene (169,0 kb) ACTC1, ACTN2, DTNA, DMD, HCN4, LDB3, LMNA, MIB1, MYBPC3, MYH7, NKX2-5, PRDM16, RYR2, SCN5A, TAFAZZIN, TNNT2, TPM1, TTN	4 - 6 Wo	E
Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen * Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Long-QT-Syndrom (LQT) * Gen-Panel: ID013.01, 18 Gene (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN	3 - 5 Wo	E
Marfan-Syndrom (MFS) *, # Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1 TGFBR1, TGFBR2	2 - 4 Wo	E
Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder * Gen-Panel ID194.03 Marfan-Syndrom und ähnliche Krankheitsbilder: 39 Gene (134,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL11A1, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MTHFR, MYH11, MYLK, PLOD1, PRDM5, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFBR1, TGFBR2 Thorakales Aortenaneurysma (TAA/D): 13 Gene (36,4 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MYH11, MYLK, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469 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
Muskelerkrankungen mit Herzbeteiligung * Gen-Panel: ID123.02 Muskelerkrankungen mit Herzbeteiligung: 34 Gene (220,8 kb) BAG3, BVES, CAV3, CRYAB, DES, DMD, DPM3, EMD, FHL1, FKRP, FKTN, FLNC, JAG2, KY, LDB3, LAMA2, LIMS2, LMNA, MYL2, MYOT, POMT1, PYROXD1, SGCA, SGCB, SGCD, SGCG, SVIL, SYNE1, SYNE2, TCAP, TMEM43, TOR1AIP1, TTN, UNC45B Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Gliedergürtelmuskeldystrophie (LGMD): 17 Gene (131,7 kb) BVES, CAV3, DES, DPM3, FKRP, FKTN, JAG2, LAMA2, LIMS2, POMT1, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TTN 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
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: ID349.00 Plötzlicher Herztod: 110 Gene (366,9 kb) ABCC9, ACTC1, ACTN2, AKAP9, ALG10B, ALPK3, ANK2, ANKRD1, BAG3, BAG5, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, EMD, EYA4, FGF12, FHL1, FHOD3, FKRP, FKTN, FLNC, GATAD1, GLA, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, NKX2-5, PKP2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RANGRF, RBM20, RPL3L, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SEMA3A, SGCD, SLC4A3, SLMAP, SNTA1, TBX5, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, VCL Arrhythmien (BRGDA, LQT) und plötzlicher Herztod: 65 Gene (268,2 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, EMD, FGF12, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LMNA, MYBPC3, MYH6, NKX2-5, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN Kardiomyopathien (HCM, DCM) und plötzlicher Herztod: 63 Gene (247,7 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, BAG5, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSG2, DSP, EMD, EYA4, FHL1, FHOD3, FKRP, FKTN, FLNC, GATAD1, GLA, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, NKX2-5, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL	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

Erkrankung/Diagnostik	Dauer	Material
Herz- und Gefäßerkrankungen		
Restriktive Kardiomyopathie (RCM) *	3 - 5 Wo	E
Gen-Panel: ID105.01, 14 Gene (31,4 kb)		
ACTC1, BAG3, DES, FLNC, KIF20A, MCM10, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR		
Short-QT-Syndrom (SQT) *	3 - 5 Wo	E
Gen-Panel: ID233.01, 7 Gene (22,1 kb)		
CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3		
Sick-Sinus-Syndrom (SSS) *	3 - 5 Wo	E
Gen-Panel: ID107.01, 4 Gene (16,5 kb)		
GNB2, HCN4, MYH6, SCN5A		
Speicherkrankheiten mit Herzbeteiligung *	3 - 5 Wo	E
Gen-Panel: ID149.01, 13 Gene (21,5 kb)		
ATP7B, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, PRKAG2, SLC40A1, TFR2, TTR		
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) *	3 - 5 Wo	E
Gen-Panel: ID020.01		
Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD): 16 Gene (41,0 kb)		
ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4		
Nicht-syndromales thorakales Aortenaneurysma (AAT): 10 Gene (22,8 kb)		
ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4		
Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb)		
SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2		
Viszerale Heterotaxie (HTX) *	3 - 5 Wo	E
Gen-Panel: ID145.01, 18 Gene (72,4 kb)		
ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, DNAAF1, DNAH5, DNAH9, DNAH11, MMP21, MNS1, NODAL, ODAD2, PKD1L1, ZIC3		
Zerebrale Kleingefäßerkrankung (BSVD) *	3 - 5 Wo	E
Gen-Panel: ID325.00, 3 Gene (12,0 kb)		
COL4A1, COL4A2, COLGALT1		
Zerebrovaskuläre Erkrankungen und Schlaganfall *	4 - 6 Wo	E
Gen-Panel: ID234.02		
Zerebrovaskuläre Erkrankungen und Schlaganfall: 44 Gene (130,0 kb)		
ACE, ADA2, ACTA2, ALOX5AP, APOE, APP, CBS, CCM2, COL3A1, COL4A1, COL4A2, COLGALT1, CST3, F2, F5, FBN1, FLNA, GAA, GLA, GSN, GUCY1A1, HTRA1, ITM2B, JAG1, KRIT1, MTHFR, MYH11, MYLK, NOS3, NOTCH3, OTC, PDCD10, POLG, PRKCH, PRNP, RNF213, SLC2A10, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, TREX1, TTR		
Zerebrale Amyloidangiopathie: 6 Gene (7,1 kb)		
APP, CST3, GSN, ITM2B, PRNP, TTR		
Zerebrale Kleingefäßerkrankung (BSVD): 3 Gene (12,0 kb)		
COL4A1, COL4A2, COLGALT1		
Zerebrale kavernöse Fehlbildung (CCM): 3 Gene (4,2 kb)		
CCM2, KRIT1, PDCD10		
Zerebrales Aneurysma und Dissektion: 10 Gene (32,8 kb)		
ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2		
Moyamoya-Erkrankung (MYMY): 4 Gene (22,2 kb)		
ACTA2, GUCY1A1, JAG1, RNF213		
CADASIL, CARASIL: 2 Gene (8,4 kb)		
HTRA1, NOTCH3		
HNO-Erkrankungen		
Branchiootorenales Syndrom (BOR) *	3 - 5 Wo	E
Gen-Panel: ID315.00, 5 Gene (10,2 kb)		
EYA1, SALL1, SIX1, SIX5, TFAP2A		
Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) *	3 - 5 Wo	E
Gen-Panel: ID170.04, 37 Gene (62,5 kb)		
ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11		
Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA) *	4 - 6 Wo	E
Gen-Panel: ID091.03, 48 Gene (144,6 kb)		
ABCC1, ACTG1, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MCM2, MYH14, MYH9, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PLS1, POU4F3, PTPRQ, REST, SCD5, SIX1, SCL12A2, SLC17A8, SLC44A4, TBC1D24, TECTA, TMC1, TNC, TRRAP, WFS1		
Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB) *	4 - 6 Wo	E
Gen-Panel: ID092.03, 75 Gene (207,8 kb)		
ADCY1, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL4A6, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MPZL2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PPIP5K2, PJVK, PNPT1, POU3F4, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN		

Erkrankung/Diagnostik	Dauer	Material
HNO-Erkrankungen		
Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik * Gen-Panel: ID237.02 Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik: 117 Gene (326,4 kb) ABCC1, ACTG1, ADCY1, AIFM1, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GIPC3, GJB2, GJB3, GJB6, GREB1L, GPRASP2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MCM2, MET, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PJVK, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, WBP2, FS1, WHRN Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA): 48 Gene (144,6 kb) ABCC1, ACTG1, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MCM2, MYH14, MYH9, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PLS1, POU4F3, PTPRQ, REST, SCD5, SIX1, SCL12A2, SLC17A8, SLC44A4, TBC1D24, TECTA, TMC1, TNC, TRRAP, WFS1 Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB): 75 Gene (207,8 kb) ADCY1, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL4A6, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MPZL2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PPIP5K2, PJVK, PNPT1, POU3F4, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX): 6 Gene (11,8 kb) AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX	4 - 6 Wo	E
Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX) * Gen-Panel: ID290.00, 7 Gene (16,8 kb) AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX	3 - 5 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
Syndromale Schwerhörigkeit, umfassende Diagnostik * Gen-Panel: ID190.01 Syndromale Schwerhörigkeit, umfassende Diagnostik: 109 Gene (317,3 kb) ABHD12, ADGRV1, AIFM1, ALMS1, ANKH, ARSG, ATP1A3, ATP6V0A4, ATP6V1B1, BCAP31, BCS1L, BRAF, BSND, CACNA1D, CATSPER2, CD151, CDH23, CHD7, CHSY1, CIB2, CISD2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, DCAF17, DIAPH3, DLX5, DNMT1, DSPP, EDN3, EDNRB, ERAL1, ESPN, EXOSC2, EYA1, FGF3, FGFR3, FOXC1, FOXI1, GATA3, GJA1, GJB2, GPSM2, HARS1, HARS2, HSD17B4, KCNE1, KCNJ10, KCNQ1, KITLG, LARS2, LHX3, LRP2, MAF, MANBA, MITF, MPZ, MYH14, MYH9, MYO7A, NLRP3, PAX3, PCDH15, PDZD7, PEX1, PEX6, PLOD3, PMP22, POLD1, POLR1C, POLR1D, PRDM5, PRPS1, PTPN11, RAF1, RPGR, RPS6KA3, SALL1, SALL4, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLC4A11, SLC52A2, SLC52A3, SNAI2, SOX10, SPATA5, TCOF1, TFAP2A, TIMM8A, TWNK, TYR, USH1C, USH1G, USH2A, WFS1, WHRN, ZNF469 Usher-Syndrom (USH): 13 Gene (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USHC, USH1G, USH2A, WHRN Stickler-Syndrom (STL): 6 Gene (21,2 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Alport-Syndrom (ATS): 4 Gene (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9 Waardenburg-Syndrom (WS): 8 Gene (9,4 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Perrault-Syndrom (PRLTS): 6 Gene (9,3 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb) BRAF, PTPN11, RAF1 CHARGE-Syndrom: 2 Gene (11,3 kb) CHD7, SEMA3E	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Immunologische Erkrankungen		
Autoinflammatorische Syndrome * Gen-Panel: ID087.04 Autoinflammatorische Syndrome: 37 Gene (61,0 kb) ADA2, AP1S3, ACP5, ARPC1B, CARD14, ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NTRK1, OTULIN, PLCG2, POMP, PSMA3, PSMB4, PSMB8, PSMB9, PSMB10, PSMG2, PSTPIP1, RBCK1, RIPK1, SLC29A3, SOCS1, STING1, TNFAIP3, TNFRSF1A, TRAF3IP2, TRNT1, WDR1 Periodische Fiebersyndrome: 12 Gene (24,4 kb) ADA2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NTRK1, OTULIN, RIPK1, TNFRSF1A, TRNT1, WDR1	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
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, RTEL1, SRP72, TERC, TERT, TP53, ZCCHC8 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8	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 Syndromale Erkrankung 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
Periodische Fiebersyndrome (HPF) * Gen-Panel: ID088.03, 7 Gene (18,1 kb) MEFV, MVK, NLRC4, NLRP12, NLRP3, PLCG2, TNFRSF1A	3 - 5 Wo	E
Lungen- und Respiratorische Erkrankungen		
Interstitielle Lungenerkrankung und Lungenfibrose * Gen-Panel: ID341.01 Interstitielle Lungenerkrankung und Lungenfibrose: 40 Gene (80,9 kb) ABCA3, ACD, AP3B1, AP3D1, COPA, CSF2RA, CSF2RB, DCLRE1B, DKC1, FAM111B, FARSA, FARSB, FLNA, FOXF1, GBA1, HPS1, HPS4, ITGA3, MARS1, NAF1, NHP2, NKX2-1, NOP10, OAS1, PARN, RPA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STING1, TERC, TERT, TINF2, WRAP53, ZCCHC8 Pulmonale Alveolarproteinose (SMDP, ILD): 10 Gene (17,7 kb) ABCA3, CSF2RA, CSF2RB, MARS1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC7A7, OAS1 Telomer-assoziierte Lungenfibrose (PFBMFT, DKC): 14 Gene (22,7 kb) ACD, DCLRE1B, DKC1, NAF1, NHP2, NOP10, PARN, RPA1, RTEL1, TERC, TERT, TINF2, WRAP53, ZCCHC8 Syndrome mit Lungenfibrose: 16 Gene (32,6 kb) ACD, AP3B1, AP3D1, DKC1, FAM111B, HPS1, HPS4, NAF1, NHP2, NOP10, PARN, RTEL1, SLC34A2, TERT, TINF2, WRAP53 Syndrome mit diffuser parenchymaler Lungenerkrankungen: 10 Gene (25,0 kb) COPA, FARSA, FARSB, FLNA, FOXF1, GBA1, ITGA, NKX2-1, SMPD1, STING1	3 - 5 Wo	E
Kongenitales zentrales Hypoventilationssyndrom (CCHS) * Gen-Panel: ID185.01, 5 Gene (9,7 kb) LBX1, MECP2, MYO1H, PHOX2B, RET	2 - 4 Wo	E
Lungenkarzinom * Gen-Panel: ID260.02, 33 Gene (122,5 kb) ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTYH, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53	4 - 6 Wo	E
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) * Gen-Panel: ID085.02, 42 Gene (132,9 kb) ARMC4, CCDC103, CCDC39, CCDC40, CCDC65, CCDC114, CCDC151, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH5, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC6, LRRC56, MCIDAS, NEK10, NME8, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC12, TTC25, ZMYND10	4 - 6 Wo	E
Pulmonale Hypertonie (PAH) * Gen-Panel: ID281.01, 23 Gene (49,9 kb) ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4	3 - 5 Wo	E
Pulmonale Surfactant-Stoffwechselstörung (SMDP) * Gen-Panel: ID168.01, 6 Gene (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 Wo	E

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

Erkrankung/Diagnostik	Dauer	Material
Mikrozephalien und Makrozephalien		
Makrozephalie, umfassende Diagnostik * Gen-Panel: ID070.03 Makrozephalie, umfassende Diagnostik: 135 Gene (364.6 kb) ABCC9, ADK, AKT1, AKT3, ALKBH8, AMER1, ANKH, APC2, ASPA, ASXL2, BRAF, BRWD3, CAMK2G, CCDC22, CCDC88C, CCND2, CDH2, CDKN1C, CHD1, CHD3, CHD8, CRADD, CSF1R, CUL4B, DIS3L2, DDX3X, DEAF1, DNMT3A, DVL1, DVL3, EED, EML1, EXT2, EZH2, FGFR3, FIBP, FMR1, FOXP1, GATAD2B, GCDH, GFAP, GLI3, GPC3, GPC4, GRIA3, GUSB, H1-4, HEPACAM, HERC1, HRAS, HSD17B4, HUWE1, IGBP1, INPPL1, KDM5C, KIF7, KPTN, KRAS, L1CAM, LAMB1, LBR, LZTR1, MAN2B1, MAP2K1, MAP2K2, MAPK1, MECP2, MED12, MITF, MLC1, MPDZ, MRAS, MSL3, MTOR, NF1, NFIB, NFIX, NONO, NRAS, NSD1, NXN, OFD1, OPHN1, PAK1, PHF21A, PIGA, PIGM, PIGN, PIGT, PIK3CA, PIK3R2, PKDCC, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, PTPN11, RAB39B, RAC1, RAF1, RIN2, RIT1, RNF125, RNF135, ROR2, RRAS2, SEC23B, SETD2, SHANK3, SHOC2, SHROOM4, SNX10, SOS1, SOS2, SPOP, SPRED1, SPRED2, STRADA, SUFU, SUZ12, TBC1D7, TCIRG1, TMCO1, TNFRSF11A, TRIO, TRIP12, UPF3B, WASHC5, WDR81, WNT5A, ZBTB7A, ZBTB20, ZBTB42, ZDHHC9 Cowden-Syndrom (CWS): 4 Gene (8,2kb) AKT1, PIK3CA, PTEN, SEC23 Robinow-Syndrom (RRS, DRS): 5 Gene (9,5 kb) DVL1, DVL3, NXN, ROR2, WNT5A Großwuchssyndrome: 15 Gene (38,9 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, FIBP, GPC3, GPC4, NFIX, NSD1, OFD1, PTEN, RNF125, SETD2, SUZ12 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Intellektuelle Entwicklungsstörungen und Makrozephalie: 49 Gene (147,9 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, H1-4, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9	4 - 6 Wo	E
Pontozerebelläre Hypoplasie (PCH) * Gen-Panel: ID071.02, 26 Gene (60,5 kb) AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34 TSEN54, VPS51, VPS53, VRK1	3 - 5 Wo	E
Primäre Mikrozephalie, autosomal-rezessiv (MCPH) * Gen-Panel: ID031.02, 25 Gene (107,1 kb) ANKLE2, ASPM, CDK6, CIT, CDK5RAP2, CENPE, CENPJ, CEP135, CEP152, COPB2, KIF14, KNL1, MAP11, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PHC1, SASS6, STIL, WDFY3, WDR62, ZNF335	4 - 6 Wo	E
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
Sotos-Syndrom (SOTOS) und ähnliche Fehlbildungssyndrome Gen-Panel: ID181.01, 4 Gene (18,8 kb) APC2, EZH2, NFIX, NSD1	3 - 5 Wo	E
Wachstumsstörung und Makrozephalie * Gen-Panel: ID072.03 Wachstumsstörung und Makrozephalie: 30 Gene (80,9 kb) AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12 Großwuchssyndrom und Makrozephalie: 11 Gene (33,0 kb) DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12 Noonan-Syndrom und Makrozephalie: 10 Gene (15,2 kb) BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS, SHOC2, SOS1	3 - 5 Wo	E
Mitochondriopathien		
Coenzym-Q10-Mangel (COQ10D) * Gen-Panel: ID225.01, 15 Gene (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2	3 - 5 Wo	E
Kombinierter Defekt der oxidativen Phosphorylierung (COXPD) * Gen-Panel: ID287.00, 51 Gene (67,7 kb) AARS2, AIFM1, ATP5F1A, C1QBP, CARS2, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL44, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NSUN3, QRSL1, PNPT1, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, 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

Erkrankung/Diagnostik	Dauer	Material
Mitochondriopathien		
Defizienz der mitochondrialen Komplexe I bis V (MCDN) *	3 - 5 Wo	E
Panel: ID074.03		
Defizienz der mitochondrialen Komplexe I bis V (MCDN): 83 Gene (60,7 kb)		
ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX5A, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, CYC1, DNAJC30, FOXRED1, LRPPRC, LYRM7*, MTFMT, NDUFA1, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, PET100, PET117, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SURF1, TACO1, TIMMDC1, TMEM70, TMEM126B, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ		
Mitochondrialer Komplex-I-Mangel, kernkodierter Typ (MC1DN): 39 Gene (29,3 kb)		
ACAD9, DNAJC30, FOXRED1, MTFMT, NDUFA1, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, TIMMDC1, TMEM126B		
Mitochondrialer Komplex-II-Mangel, kernkodierter Typ (MC2DN): 4 Gene (3,7 kb)		
SDHA, SDHAF, SDHB, SDHD		
Mitochondrialer Komplex-III-Mangel, kernkodierter Typ (MC3DN): 10 Gene (7,1 kb)		
BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1		
Mitochondrialer Komplex-IV-Mangel, kernkodierter Typ (MC4DN): 23 Gene (15,8 kb)		
COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, COX5A, LRPPRC, NDUFA4, PET100, PET117, SCO1, SCO2, SURF1, TACO1		
Mitochondrialer Komplex-V-(ATP-Synthase-)Mangel, kernkodierter Typ (MC5DN): 7 Gene (4,8 kb)		
ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, TMEM70		
Mitochondriales DNA-Depletionssyndrom (MTDPS) *	3 - 5 Wo	E
Gen-Panel: ID324.00, 19 Gene (25,6 kb)		
AGK, DGUOK, FBXL4, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP		
Mitochondriale Myopathie / Enzephalopathie #	3 - 5 Wo	E
Gen-Panel: ID702.00, 27 Gene (7,0 kb)		
MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND2, MT-ND5, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TW		
Mitochondriengenom #	3 - 5 Wo	E
Gen-Panel: ID703.00, 37 Gene (11,4 kb)		
MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY		
Nukleär-kodierte mitochondriale Erkrankungen *	4 - 6 Wo	E
Gen-Panel: ID704.01, 312 Gene (406,1 kb)		
AARS2, ABCB7, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH4A1, ALDH6A1, AMACR, AMT, ANO10, APTX, ATL1, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATPAF2, AUH, BCAT2, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C1QBP, CARS2, CISD2, COA3, COA5, COA6, COA8, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX5A, COX6A2, COX6B1, CPS1, CPT1A, CPT2, CYB5R3, CYC1, CYCS, CYP27A1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, EARS2, ELAC2, ERCC6, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBP1, FBXL4, FH, FOXRED1, FXN, GAMT, GARS1, GATB, GATC, GATM, GCDH, GCK, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GTPBP3, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HK1, HLCS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSPD1, HTRA2, IDH1, IDH2, IDH3B, ISCU, IVD, KARS1, KIF1B, KIF5A, L2HGDH, LARS2, LIAS, LRPPRC, LYRM4, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICOS13, MIEF2, MIPEP, MLYCD, MMAA, MMAB, MMADHC, MMUT, MPV17, MRM2, MRPL12, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS28, MRPS34, MRPS7, MTFMT, MTO1, MTPAP, MTRFR, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NSUN3, NUBPL, OAT, OPA1, OPA3, OTC, OXCT1, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PET117, PINK1, PNKD, PNPLA2, PNPT1, POLG, POLG2, PPOX, PRKN, PTCD3, PUS1, QRSL1, RARS2, REEP1, RMND1, RMRP, RNASEH1, RRM2B, SACS, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A10, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC33A1, SLC6A8, SPART, SPAST, SPG7, STAR, SUCLA2, SUCLG1, SURF1, TACO1, TAFAZZIN, TARS2, TFAM, TIMM22, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UNG, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, VARS2, WFS1, WWOX, XPNPEP3, YARS2, YWHAE		
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB) *	3 - 5 Wo	E
Gen-Panel: ID300.00		
Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB): 10 Gene (17,9 kb)		
DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK		
Progressive externe Ophthalmoplegie, autosomal-dominant (PEOA): 6 Gene (12,4 kb)		
DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK		
Progressive externe Ophthalmoplegie, autosomal-rezessiv (PEOB): 5 Gene (9,2 kb)		
DGUOK, POLG, RNASEH1, TK2, TOP3A		

Erkrankung/Diagnostik	Dauer	Material
Multisystem-Fehlbildungssyndrome		
Alport-Syndrom (ATS) *	3 - 5 Wo	E
Gen-Panel: ID099.00, 4 Gene (21,0 kb)		
COL4A3, COL4A4, COL4A5, MYH9		
Bardet-Biedl-Syndrom (BBS) *	3 - 5 Wo	E
Gen-Panel: ID093.02, 21 Gene (39,0 kb)		
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
CHARGE-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID307.00, 3 Gene (12,9 kb)		
CHD7, SEMA3E, TBX22		
Coffin-Siris-Syndrom (CSS) *	3 - 5 Wo	E
Gen-Panel: ID118.01, 10 Gene (34,2 kb)		
ARID1A, ARID1B, ARID2, DPF2, SMARCC2, SMARCA4, SMARCB1, SMARCE1, SOX4, SOX11		
Cornelia-de-Lange-Syndrom (CDLS) *	3 - 5 Wo	E
Gen-Panel: ID033.02, 8 Gene (32,0 kb)		
ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS		
Fehlbildungssyndrome mit überwiegend fazialer Beteiligung *	3 - 5 Wo	E
Gen-Panel: ID279.00		
Fehlbildungssyndrome mit überwiegend fazialer Beteiligung: 25 Gene (90,9 kb)		
C2CD3, COL11A2, CPLANE1, DDX59, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GRIP1, IFT57, INTU, KIAA0753, MEGF8, MYH3, MYMK, MYT1, OFD1, RAB23, RBM10, TCTN3, TGDS, TMEM107, TNNI2, TWIST1		
Akrozephalosyndaktylie-Syndrom (ACS): 6 Gene (17,0 kb)		
FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1		
Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Fraser-Syndrom (FRASRS): 3 Gene (24,8 kb)		
FRAS1, FREM2, GRIP1		
Pierre-Robin-Syndrom: 4 Gene (9,7 kb)		
COL11A2, MYMK, RBM10, TGDS		
FG-Syndrom (FGS) *	3 - 5 Wo	E
Gen-Panel: ID215.00, 3 Gene (17,2 kb)		
CASK, FLNA, MED12		
Kabuki-Syndrom (KABUK) *, #	3 - 5 Wo	E
Gen-Panel: ID127.00, 2 Gene (20,8 kb)		
KDM6A, KMT2D		
Marfan-Syndrom (MFS) *, #	2 - 4 Wo	E
Gen-Panel: ID022.00, 3 Gene (11,8 kb)		
FBN1 TGFBR1, TGFBR2		
Noonan-Syndrom (NS) *	3 - 5 Wo	E
Gen-Panel: ID023.06, 16 Gene (27,4 kb)		
BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2		
Orofaziodigitales Syndrom (OFD) *	3 - 5 Wo	E
Gen-Panel: ID265.00, 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
RASopathien *	3 - 5 Wo	E
Gen-Panel: ID015.05		
RASopathien: 21 Gene (40,1 kb)		
BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2		
Noonan-Syndrom (NS): 15 Gene (24,7 kb)		
BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2		
Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb)		
BRAF, KRAS, MAP2K1, MAP2K2		
LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb)		
BRAF, PTPN11, RAF1		
Rubinstein-Taybi-Syndrom (RSTS) *	3 - 5 Wo	E
Gen-Panel: ID142.01, 3 Gene (24,3 kb)		
CREBBP, EP300, SRCAP		
Stickler-Syndrom (STL) *	3 - 5 Wo	E
Gen-Panel: ID062.00, 6 Gene (21,2 kb)		
COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3		
Tuberöse Sklerose (TSC) *	2 - 4 Wo	E
Gen-Panel: ID332.00, 2 Gene (8,9 kb)		
TSC1, TSC2		
VACTERL-Assoziation *	3 - 5 Wo	E
Gen-Panel: ID258.02, 27 Gene (72,8 kb)		
BRCA2, CHD7, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGF8, FOXF1, GLI3, HAAO, HOXD13, HSPA6, MNX1, RECQL4, SALL1, KYNU, MYCN, NADSYN1, RAD51C, TRAP1, WBP11, ZIC3		
Weill-Marchesani-Syndrom (WMS) *	3 - 5 Wo	E
Gen-Panel: ID230.00, 4 Gene (20,7 kb)		
ADAMTS10, ADAMTS17, FBN1, LTBP2		
Zellweger-Syndrom (ZWS) *	3 - 5 Wo	E
Gen-Panel: ID084.00, 14 Gene (22,3 kb)		
ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
Arthrogrypose * Gen-Panel: ID200.01 Arthrogrypose: 61 Gene (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNM2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2 Arthrogryposis multiplex congenita (AMC): 6 Gene (58,2 kb) ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1 Distale Arthrogrypose (DA): 11 Gene (40,3 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1 Letales kongenitales Kontraktursyndrom (LCCS): 11 Gene (31,5 kb) ADCY6, ADGRG6, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42 Fetale Akinesie-Deformation-Sequenz (FADS): 4 Gene (7,6 kb) DOK7, MUSK, NUP88, RAPSN	4 - 6 Wo	E
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT, HMSN) * Gen-Panel: ID052.02 Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT, HMSN): 41 Gene (98,5 kb) AARS1, AIFM1, ATP1A1, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PRPS1, RAB7A, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth-Neuropathie, axonal, autosomal-dominant (CMT2): 22 Gene (60,2 kb) AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, axonal, autosomal-rezessiv (CMT2): 10 Gene (25,1 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MME, MPV17, SPG11, TRIM2 Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46	4 - 6 Wo	E
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT, HMSN) * Gen-Panel: ID051.02 Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT, HMNS): 26 Gene (56,7 kb) CNTNAP1, COX6A1, DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-dominant (CMT1): 7 Gene (6,3 kb) EGR2, GDAP1, LITAF, MPZ, NEFL, 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: ID312.01 Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik: 62 Gene (146,3 kb) AARS1, AIFM1, ARHGEF10, ATP1A1, CNTNAP1, COX6A1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SORD, SPG11, SPTLC1, SURF1, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, dominant (CMT1): 7 Gene (6,3 kb) EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, rezessiv (CMT4): 12 Gene (33,8 kb) EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2): 29 Gene (79,8 kb) AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, RAB7A, 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 Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46 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	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
Distale Arthrogrypose (DA) *	3 - 5 Wo	E
Gen-Panel: ID196.02, 11 Gene (40,5 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Distale motorische Neuronopathie (HMN, DSMA) *	3 - 5 Wo	E
Gen-Panel: ID254.00		
Distale motorische Neuronopathie (HMN, DSMA): 17 Gene (38,7 kb)		
ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4, WARS1		
Distale motorische Neuronopathie, dominant (HMN, DHMN): 12 Gene (26,7 kb)		
BSCL2, DCTN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, TRPV4, WARS1		
Distale motorische Neuronopathie, rezessiv (DSMA): 5 Gene (12,0 kb)		
ATP7A, DNAJB2, IGHMBP2, PLEGHG5, SIGMAR1		
Distale Myopathie (MPD) *	4 - 6 Wo	E
Gen-Panel: ID328.00, 18 Gene (169,4 kb)		
ACTN2, ADSS1, ANO5, CAV3, CRYAB, DES, DNAJB6, DYSF, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, NEB, TIA1, TCAP, TTN		
Emery-Dreifuss-Muskeldystrophie (EDMD) *	3 - 5 Wo	E
Gen-Panel: ID121.00, 6 Gene (51,8 kb)		
EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43		
Fetale Akinesie-Sequenz (FADS) *	3 - 5 Wo	E
Gen-Panel: ID201.00, 10 Gene (30,3 kb)		
CHRNA1, CHRND, CHRNG, DOK7, GBE1, MUSK, MYOD1, NUP88, RAPSN, RYR1		
Gliedergürtelmuskeldystrophie (LGMD) *	4 - 6 Wo	E
Gen-Panel: ID122.04		
Gliedergürtelmuskeldystrophie (LGMD): 37 Gene (196,8 kb)		
ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DNAJB6, DPM3, DYSF, FKRP, FKTN, GMPPB, HNRNPDL, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN		
Gliedergürtelmuskeldystrophie, autosomal-rezessiv (LGMDR): 34 Gene (191,8 kb)		
ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DPM3, DYSF, FKRP, FKTN, GMPPB, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TRAPPC11, TRIM32, TTN		
Gliedergürtelmuskeldystrophie, autosomal-dominant (LGMDD): 7 Gene (23,2 kb)		
CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3		
Gliedergürtelmuskeldystrophie-Dystroglykanopathie (MDDGC): 11 Gene (17,6 kb)		
CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2		
Ullrich-Muskeldystrophie (UCMD): 3 Gene (15,7 kb)		
COL6A1, COL6A2, COL6A3		
Bethlem-Myopathie (BTHLM): 3 Gene (15,7 kb)		
COL6A1, COL6A2, COL6A3		
Kongenitales myasthenes Syndrom (CMS) *	3 - 5 Wo	E
Gen-Panel: ID130.00		
Kongenitales myasthenes Syndrom (CMS): 25 Gene (55,9 kb)		
AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, MYO9A, PREPL, RAPSN, SCN4A, SLC5A7, SLC18A3, SLC25A1, SNAP25, SYT2, VAMP1		
Kongenitales myasthenes Syndrom (CMS), präsynaptisch: 8 Gene (21,9 kb)		
AGRN, CHAT, MYO9A, SLC5A7, SLC18A3, SLC25A1, SYT2, VAMP1		
Kongenitales myasthenes Syndrom (CMS), synaptisch oder postsynaptisch: 18 Gene (40,1 kb)		
AGRN, ALG2, ALG14, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, PREPL, RAPSN, SCN4A, SNAP25		
Kongenitale Strukturmyopathie *	3 - 5 Wo	E
Gen-Panel: ID212.01		
Kongenitale Strukturmyopathie: 20 Gene (78,8 kb)		
ACTA1, BIN1, CCDC78, CFL2, DNM2, KBTBD13, KLHL40, KLHL41, LMOD2, MAP3K20, MTM1, MTMR14, MYPN, NEB, RYR1, SELENON, SPEG, TNNT1, TPM2, TPM3		
Core-Myopathie: 4 Gene (18,9 kb)		
ACTA1, RYR1, SELENON, TPM3		
Nemalin-Myopathie (NEM): 11 Gene (40,6 kb)		
ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, TNNT1, TPM2, TPM3, MYPN, NEB		
Zentronukleäre Myopathie (CNM): 7 Gene (21,4 kb)		
Letales kongenitales Kontraktursyndrom (LCCS) *	3 - 5 Wo	E
Gen-Panel: ID197.00, 12 Gene (34,6 kb)		
ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42		
Muskelerkrankungen mit Herzbeteiligung *	4 - 6 Wo	E
Gen-Panel: ID123.02		
Muskelerkrankungen mit Herzbeteiligung: 34 Gene (220,8 kb)		
BAG3, BVES, CAV3, CRYAB, DES, DMD, DPM3, EMD, FHL1, FKRP, FKTN, FLNC, JAG2, KY, LDB3, LAMA2, LIMS2, LMNA, MYL2, MYOT, POMT1, PYROXD1, SGCA, SGCB, SGCD, SGCG, SVIL, SYNE1, SYNE2, TCAP, TMEM43, TOR1AIP1, TTN, UNC45B		
Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb)		
BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B		
Gliedergürtelmuskeldystrophie (LGMD): 17 Gene (131,7 kb)		
BVES, CAV3, DES, DPM3, FKRP, FKTN, JAG2, LAMA2, LIMS2, POMT1, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TTN		
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		

Erkrankung/Diagnostik	Dauer	Material
Muskelerkrankungen		
Muskelerkrankungen, umfassende Diagnostik * Gen-Panel: ID336.00 Muskelerkrankungen, umfassende Diagnostik: 246 Gene (749,8 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, AGRN, ALDOA, ALG14, ALG2, AMPD1, ANO5, ASAH1, ASCC1, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNA2, DNAJB6, DNM2, DNMT3B, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFPT1, GMPPB, GNE, GYG1, GYS1, HADH, HADHA, HADHB, HINT1, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB8, IGHMBP2, INPP5K, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIG3, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MAP3K20, MATR3, MEGF10, MFN2, MGME1, MICU1, MPV17, MPZ, MTM1, MTMR14, MTRFR, MUSK, MYBPC1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, NEFL, OPA1, ORAI1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SCO2, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC52A3, SLC5A7, SMCHD1, SMN1, SNAP25, SPEG, SPG7, SPG11, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TCAP, TFAM, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TRMT5, TRPV4, TSFM, TTN, TUBB3, TWNK, TYMP, UBA1, UNC45B, VAMP1, VCP, VMA21, VRK1, YARS2 Kongenitale, distale und metabolische Myopathien: 110 Gene (382,6 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, ALDOA, AMPD1, ANO5, BAG3, BIN1, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, ECEL1, ENO3, ETFA, ETFB, ETFDH, FLAD1, FLNC, FXR1, GAA, GBE1, GNE, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LDB3, LAMP2, LDHA, LIG3, LMOD3, LPIN1, MAP3K20, MATR3, MEGF10, MGME1, MTM1, MTMR14, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYMK, MYOT, MYPN, NEB, TNNI2, OPA1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, PYROXD1, RBCK1, RRM2B, RYR1, SELENON, SLC22A5, SLC25A20, SLC25A4, SPEG, STAC3, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TIA1, TK2, TNNT1, TNNT3, TPM2, TPM3, TTN, TYMP, UBA1, YARS2 Gliedergürtelmuskeldystrophie (LGMD) und weitere Muskeldystrophien: 47 Gene (270,0 kb) ANO5, B3GALNT2, B4GAT1, BVES, CAPN3, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, HNRNPDL, LAMA2, LARGE1, LIMS2, LMNA, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RXYLT1, SGCA, SGCB, SGC, SGCG, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	4 - 6 Wo	E
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
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
Sensorisch-autonome Neuropathie (HSAN, HSN) * Gen-Panel: ID086.01, 15 Gene (52,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, WNK1	3 - 5 Wo	E
Spinale Muskelatrophie (SMA) * [1] Gen-Panel: ID152.01 Spinale Muskelatrophie (SMA): 31 Gene (69,5 kb) ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS1 Proximale spinale Muskelatrophie (SMA): 10 Gene (26,6 kb) ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, SMN1, SMN2, TRIP4, UBA1, VAPB Distale spinale Muskelatrophie (DSMA, HMN): 18 Gene (41,9 kb) ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, SIGMAR1, PLEKHG5, REEP1, SETX, SLC5A7, TRPV4, UBA1, WARS1 [1] Es erfolgt eine Stufendiagnostik bei SMN1 und SMN2 gemäß EBM Kapitel 11.4.2.	3 - 5 Wo	E
Walker-Warburg-Syndrom (WWS, MDDGA) * Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Zentronukleäre Myopathie (CNM) * Gen-Panel: ID257.00, 7 Gene (21,4 kb) CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Alzheimer-Krankheit (AD) *	3 - 5 Wo	E
Gen-Panel: ID157.01, 7 Gene (16,8 kb)		
ABCA7, ADAM10, APOE, APP, PSEN1, PSEN2, TF		
Amyotrophe Lateralsklerose (ALS) *	3 - 5 Wo	E
Gen-Panel: ID209.02		
Amyotrophe Lateralsklerose (ALS): 31 Gene (71,4 kb)		
ALS2, ANG, ANXA11, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP		
ALS und Frontotemporale Demenz (FTDALS): 13 Gene (21,2 kb)		
CCNF, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP		
ALS, juvenile Form: 4 Gene (21,0 kb)		
ALS2, SETX, SIGMAR1, SPG11		
Basalganglien-Kalzifikation (IBGC) *	3 - 5 Wo	E
Gen-Panel: ID327.00, 6 Gene (11,2 kb)		
JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1		
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT, HMSN) *	3 - 5 Wo	E
Gen-Panel: ID052.02		
Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT, HMSN): 41 Gene (98,5 kb)		
AARS1, AIFM1, ATP1A1, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PRPS1, RAB7A, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1		
Charcot-Marie-Tooth-Neuropathie, axonal, autosomal-dominant (CMT2): 22 Gene (60,2 kb)		
AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, TRPV4, VCP		
Charcot-Marie-Tooth-Neuropathie, axonal, autosomal-rezessiv (CMT2): 10 Gene (25,1 kb)		
GDAP1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MME, MPV17, SPG11, TRIM2		
Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb)		
COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1		
Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb)		
AIFM1, GJB1, PDK3, PRPS1		
Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6): 3 Gene (4,5 kb)		
MFN2, PDXK, SLC25A46		
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT, HMSN) *	3 - 5 Wo	E
Gen-Panel: ID051.02		
Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT, HMSN): 26 Gene (56,7 kb)		
CNTNAP1, COX6A1, DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-dominant (CMT1): 7 Gene (6,3 kb)		
EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-rezessiv (CMT4): 12 Gene (33,8 kb)		
EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1		
Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb)		
COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1		
Hypertrophe Dejerine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb)		
EGR2, MPZ, PMP22, PRX		
Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb)		
EGR2, MPZ, CNTNAP1		
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID312.01		
Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik: 62 Gene (146,3 kb)		
AARS1, AIFM1, ARHGEF10, ATP1A1, CNTNAP1, COX6A1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SORD, SPG11, SPTLC1, SURF1, TRIM2, TRPV4, VCP, YARS1		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend, dominant (CMT1): 7 Gene (6,3 kb)		
EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22		
Charcot-Marie-Tooth-Neuropathie, demyelinisierend, rezessiv (CMT4): 12 Gene (33,8 kb)		
EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1		
Charcot-Marie-Tooth-Neuropathie, axonal (CMT2): 29 Gene (79,8 kb)		
AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, RAB7A, 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		
Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6): 3 Gene (4,5 kb)		
MFN2, PDXK, SLC25A46		
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		

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
CADASIL und CARASIL *, # Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E
Choreatiforme Bewegungsstörungen * Gen-Panel: ID272.01, 17 Gene (40,6 kb) ADCY5, ATP1A2, ATP1A3, FRRS1L, GNAO1, HPRT1, KCNMA1, NKX2-1, PDE2A, PDE10A, PNKD, PRNP, PRRT2, RNF216, SLC2A1, VPS13A, XK	3 - 5 Wo	E
Distale motorische Neuronopathie (HMN, DSMA) * Gen-Panel: ID254.00 Distale motorische Neuronopathie (HMN, DSMA): 17 Gene (38,7 kb) ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4, WARS1 Distale motorische Neuronopathei, dominant (HMN, DHMN): ID254.00, 11 Gene (18,6 kb) BSCL2, DCTN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, TRPV4, WARS1 Distale motorische Neuronopathie, rezessiv (DSMA): 5 Gene (12,0 kb) ATP7A, DNAJB2, IGHMBP2, PLEGHG5, SIGMAR1	3 - 5 Wo	E
Dystonie (DYT) * Gen-Panel: ID128.03 Dystonie (DYT): 25 Gene (55,9 kb) ACTB, ADCY5, ANO3, AOPEP, ATP1A3, COL6A3, ECHS1, GCH1, GNAL, HPCA, KCTD17, KMT2B, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VPS16 Dystonie (DYT), isolierte Form: 9 Gene (27,8 kb) ANO3, AOPEP, COL6A3, GNAL, HPCA, KMT2B, THAP1, TOR1A, TUBB4A Dystonie (DYT), kombinierte Form: 16 Gene (28,1 kb) ACTB, ADCY5, ATP1A3, ECHS1, GCH1, KCTD17, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH; VPS16	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
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: ID310.00 Frontotemporale Demenz (FTD): 18 Gene (27,9 kb) CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporale Demenz und Amyotrophe Lateralsklerose (FTDALS): 13 Gene (21,2 kb) CCNF, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporale Demenz, Einschlusskörperchenmyopathie und Paget-Syndrom (IBMPFD): 3 Gene (4,6 kb) HNRNPA1, HNRNPA2B1, 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: ID276.01 Hereditäre Ataxien, umfassende Diagnostik: 114 Gene (334,2 kb) ABCB7, ABHD12, ACO2, AFG3L2, AIFM1, ANO10, APTX, ATCAY, ATG5, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CASK, CCDC88C, CHP1, CLCN2, CLN5, COA7, 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, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PLD3, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRKCG, PRPS1, PTF1A, PUM1, RNF216, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1 Episodische Ataxie (EA): 6 Gene (20,5 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3 Spastische Ataxie (SPAX): 6 Gene (24,7 kb) AFG3L2, CHP1, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1 Spinozerebelläre Ataxie (SCA, SCAR): 46 Gene (161,8 kb) AFG3L2, ANO10, ATG5, CACNA1A, CACNA1G, CCDC88C, COQ8A, CWF19L1, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GDAP2, GRID2, GRM1, ITPR1, KCNC3, KCND3, MME, PDYN, PLD3, PMPCA, PRKCG, PUM1, RUBCN, SCYL1, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TTBK2, TBA5, VPS13D, VWA3B, WWOX, XRCC1 Spinozerebelläre Ataxie mit axonaler Neuropathie (SCAN): 3 Gene (10,6 kb) COA7, SETX, TDP1 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

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Hirnatrophie und demyelinisierende Erkrankungen des Gehirns * Gen-Panel: ID278.00 Hirnatrophie und demyelinisierende Erkrankungen des Gehirns: 55 Gene (96,0 kb) AIMP1, AIMP2, ASPA, B3GALNT2, B4GAT1, CNP, COL4A1, CRPPA, DAG1, DARS1, DEGS1, EPRS1, FAM126A, FARSA, FARSB, FKRP, FKTN, GFAP, GJC2, GMPPB, GRM7, HIKESHI, HSPD1, LARGE1, MAPT, MAT1A, MED17, MTHFS, PLP1, POLR1C, POLR3A, , POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSEN1, PYCR2, RARS1, RXYLT1, SLC25A12, SOX10, TBCD, TMEM106B, TMEM63A, TRAPPC12, TRAPPC4, TRAPPC6B, UBTF, UFM1, VPS11 Hirnatrophie: 13 Gene (24,2 kb) EXOC7, EXOC8, FARSA, FARSB, GRM7, MAPT, MED17, PSEN1, TBCD, TRAPPC4, TRAPPC6B, TRAPPC12, UBTF Hypo- und Demyelinisierung des Gehirns: 27 Gene (43,0 kb) AIMP1, AIMP2, ASPA, CNP, DARS1, DEGS1, EPRS1, FAM126A, GFAP, GJC2, HIKESHI, HSPD1, MAT1A, MTHFS, PLP1, POLR1C, POLR3A, POLR3B, PYCR2, RARS1, SLC25A12, SOX10, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Walker-Warburg-Syndrom (MDDGA): 14 Gene (23,6 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
Hyperekplexie (HKPX) * Gen-Panel: ID216.00, 9 Gene (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 Wo	E
Hypomyelinisierende Leukodystrophie (HLD) * Gen-Panel: ID277.00, 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11	3 - 5 Wo	E
Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik * Gen-Panel: ID204.04 Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik: 122 Gene (208,5 kb) AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, AIFM1, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, AUH, BOLA3, CLCN2, CLDN11, CNP, COA7, COA8, COL4A1, COL4A2, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS1, DARS2, DEGS1, EARS2, EIF2AK1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FA2H, FAM126A, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GCDH, GFAP, GFM1, GJC2, GLB1, HEPACAM, HIKESHI, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, ISCA1, ISCA2, KARS1, KCNT1, L2HGDH, LMNB1, LYRM7, MARS2, MCOLN1, MLC1, MTFMT, NAXD, NAXE, NFU1, NKX6-2, NOTCH3, NUBPL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PLAA, PLEKHG2, PLP1, PMPCB, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SDHAF1, SLC13A3, SLC16A2, SLC17A5, SOX10, SPTAN1, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYMP, TYROBP, UFM1, VPS11, ZFYVE26 Leukodystrophie mit Hypomyelinisierung (HLD): 22 Gene (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Leukodystrophie mit Peroxisomenbiogenese-Störung (PBD): 15 Gene (20,8 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH Orthochromatische Leukodystrophie: 10 Gene (14,6 kb) ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1 Metachromatische Leukodystrophie: 3 Gene (4,2 kb) ARSA, PSAP, SUMF1 Aicardi-Goutières-Syndrom (AGS): 7 Gene (11,9 kb) ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1 CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3	4 - 6 Wo	E
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, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDFT1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNPK, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAPA1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	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
Parkinson-Krankheit (PARK) * Gen-Panel: ID077.02 Parkinson-Syndrom (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

Erkrankung/Diagnostik	Dauer	Material
Neurodegenerative Erkrankungen		
Paroxysmale Dyskinesie *	3 - 5 Wo	E
Gen-Panel: ID286.00, 14 Gene (38,9 kb)		
ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24		
Sensorisch-autonome Neuropathie (HSAN, HSN) *	3 - 5 Wo	E
Gen-Panel: ID086.01, 15 Gene (52,5 kb)		
ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, WNK1		
Spastische Ataxie (SPAX) *	3 - 5 Wo	E
Gen-Panel: ID228.00, 12 Gene (34,8 kb)		
AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1		
Spastische Paraplegie (HSP, SPG) *	4 - 6 Wo	E
Gen-Panel: ID148.03		
Spastische Paraplegie (HSP, SPG): 58 Gene (117,9 kb)		
ALDH18A1, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, L1CAM, MAG, MTRFR, NIPA1, NT5C2, PCYT2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, TECPR2, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-dominant: 17 Gene (33,9 kb)		
ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, UBAP1, WASHC5, ZFYVE27		
Spastische Paraplegie (SPG), autosomal-rezessiv: 44 Gene (92,3 kb)		
AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NT5C2, PCYT2, PLP1, PNPLA6, REEP2, SELENOI, SPART, SPG7, SPG11, SPG21, TECPR2, TFG, UCHL1, VPS37A, ZFYVE26		
Spastische Paraplegie (SPG), X-chromosomal: 2 Gene (4,6 kb)		
L1CAM, PLP1		
Spinale Muskelatrophie (SMA) *	3 - 5 Wo	E
Gen-Panel: ID152.01		
Spinale Muskelatrophie (SMA): 31 Gene (69,5 kb)		
ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS1		
Proximale spinale Muskelatrophie (SMA): 10 Gene (26,6 kb)		
ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, SMN1, SMN2, TRIP4, UBA1, VAPB		
Distale spinale Muskelatrophie (DSMA, HMN): 18 Gene (41,9 kb)		
ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, SIGMAR1, PLEKHG5, REEP1, SETX, SLC5A7, TRPV4, UBA1, WARS1		
Störung der Peroxisomenbiogenese (PBD) *	3 - 5 Wo	E
Gen-Panel: ID083.01		
Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Neonatale Adrenoleukodystrophie / Infantiles Refsum-Syndrom (PBD, Typ B): 11 Gene (17,0 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26		
Heimler-Syndrom (PBD, Typ C): 2 Gene (6,8 kb)		
PEX1, PEX6		
Tuberöse Sklerose (TSC) *	2 - 4 Wo	E
Gen-Panel: ID332.00, 2 Gene (8,9 kb)		
TSC1, TSC2		
Zerebelläre Ataxie, autosomal-dominant *	3 - 5 Wo	E
Gen-Panel: ID236.02, 29 Gene (95,6 kb)		
AFG3L2, ATP1A3, CACNA1A, CACNA1G, CCDC88C, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, PDYN, PLD3, PRKCG, PUM1, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2, TUBB4A		
Zerebelläre Ataxie, autosomal-rezessiv *	4 - 6 Wo	E
Gen-Panel: ID213.03		
Zerebelläre Ataxie, autosomal-rezessiv: 50 Gene (164,7 kb)		
ANO10, APTX, ATCAY, ATG5, ATM, ATP8A2, CA8, COA7, COQ8A, CWF19L1, CYP27A1, FXN, GDAP2, GRID2, GRM1, PEX7, PHYH, PIK3R5, PMPCA, PNKP, PNPLA6, POLG, RNF216, RUBCN, SCYL1, SACS, SETX, SIL1, SLC52A2, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TTPA, TWNK, UBA5, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1		
Spinozerebelläre Ataxie (SCAR): 24 Gene (87,8 kb)		
ANO10, ATG5, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PMPCA, RUBCN, SCYL1, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP2, THG1L, TPP1, UBA5, VPS13D, VWA3B, WWOX, XRCC1		
Spinozerebelläre Ataxie mit axonaler Neuropathie (SCAN): 3 Gene (10,6 kb)		
COA7, SETX, TDP1		
Zerebelläre Ataxie mit okulomotorischer Apraxie (AOA): 4 Gene (13,3 kb)		
APTX, PIK3R5, PNKP, SETX		
Zerebelläre Ataxie mit mentaler Retardierung (CAMRQ): 4 Gene (12,9 kb)		
ATP8A2, CA8, VLDLR, WDR81		
Zerebelläre Ataxie, X-chromosomal *	3 - 5 Wo	E
Gen-Panel: ID273.00, 7 Gene (15,9 kb)		
ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6		

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Alport-Syndrom (ATS) *	3 - 5 Wo	E
Gen-Panel: ID099.00, 4 Gene (21,0 kb)		
COL4A3, COL4A4, COL4A5, MYH9		
Atypisches hämolytisch-urämisches Syndrom (AHUS) *	3 - 5 Wo	E
Gen-Panel: ID163.04, 20 Gene (42,1 kb)		
ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN		
Bardet-Biedl-Syndrom (BBS) *	3 - 5 Wo	E
Gen-Panel: ID093.02, 21 Gene (39,0 kb)		
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Bartter-Syndrom (BARTS) *	3 - 5 Wo	E
Gen-Panel: ID156.01, 8 Gene (17,7 kb)		
BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3		
Branchiootorenales Syndrom (BOR) *	3 - 5 Wo	E
Gen-Panel: ID315.00, 5 Gene (10,2 kb)		
EYA1, SALL1, SIX1, SIX5, TFAP2A		
Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen *	3 - 5 Wo	E
Gen-Panel: ID706.01, 16 Gene (30,1 kb)		
ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1		
Diabetes insipidus *	3 - 5 Wo	E
Gen-Panel: ID322.00, 5 Gene (8,4 kb)		
AQP2, AVP, AVPR2, SLC12A1, WFS1		
Fokal-segmentale Glomerulosklerose (FSGS) und Nephrotisches Syndrom (SRNS, NPHS) *	4 - 6 Wo	E
Gen-Panel: ID098.05		
Fokal-segmentale Glomerulosklerose (FSGS) und Nephrotisches Syndrom (SRNS, NPHS): 40 Gene (126,0 kb)		
ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ8B, CRB2, DAAM2, DGKE, EMP2, INF2, KANK2, KIRREL1, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PAX2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, TRPC6, WT1		
Nephrotisches Syndrom (SRNS, NPHS): 30 Gene (100,6 kb)		
ARHGAP24, ARHGDIA, AVIL, COQ8B, COL4A3, COL4A4, COL4A5, DAAM2, DGKE, EMP2, KANK2, KIRREL1, LAMA5, LAMB2, PTPRO, MAGI2, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PLCE1, SGPL1, SMARCAL1, TBC1D8B, WT1		
Fokal-segmentale Glomerulosklerose (FSGS): 13 Gene (40,6 kb)		
ACTN4, ANLN, APOL1, COL4A3, COL4A4, COL4A5, CRB2, CD2AP, INF2, LMX1B, MYO1E, PAX2, TRPC6		
Fraser-Syndrom (FRASRS) *	3 - 5 Wo	E
Gen-Panel: ID317.00, 3 Gene (24,8 kb)		
FRAS1, FREM2, GRIP1		
Galloway-Mowat-Syndrom (GAMOS) *	3 - 5 Wo	E
Gen-Panel: ID251.01, 10 Gene (12,5 kb)		
GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC		
Glomerulonephritis *	3 - 5 Wo	E
Gen-Panel: ID103.01		
Glomerulonephritis: 18 Gene (36,9 kb)		
C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD		
C3-Glomerulopathie (C3G): 3 Gene (7,2 kb)		
CFI, CFH, CFHR5		
C1q-Mangel (C1QD): 3 Gene (2,3 kb)		
C1QA, C1QB, C1QC		
Hyperoxalurie	3 - 5 Wo	E
Gen-Panel: ID363.00		
Hyperoxalurie: 6 Gene (8,6 kb)		
AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6		
Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb)		
AGXT, GRHPR, HOGA1		
Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb)		
OXGR1, SLC26A1, SLC26A6		
Hypomagnesiämie (HOMG) *	3 - 5 Wo	E
Gen-Panel ID054.02: 14 Gene (30,9 kb)		
ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6		
Hypophosphatasie, Hypophosphatämie und Rachitis *	3 - 5 Wo	E
Gen-Panel: ID269.03		
Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb)		
ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR		
Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb)		
CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3		
Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb)		
CYP2R1, CYP3A4, CYP27B1, VDR		
Hypophosphatasie (HPP): 1 Gen (1,6 kb)		
ALPL		

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Joubert-Syndrom (JBTS) * Gen-Panel: ID028.03, 40 Gene (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGAGRAM1, TTC21B, ZNF423	4 - 6 Wo	E
Kongenitale Anomalien der Niere und ableitenden Harnwege (CAKUT) * Gen-Panel: ID229.03 Kongenitale Anomalien der Niere und ableitenden Hernwege (CAKUT): 62 Gene (198,3 kb) ACE, ACTG2, AGT, AGTR1, ANOS1, BICC1, BMP4, BNC2, CDC5L, CEP55, CHD1L, CHRM3, CRKL, DSTYK, EYA1, FAT4, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3, GFRA1, GREB1L, GRIP1, HNF1B, HPSE2, ITGA8, KIF14, LIFR, LMOD1, LRIG2, LRP4, MUC1, MYH11, MYL9, MYLK, NEK8, NPHP3, NRIP1, PAX2, PBX1, REN, RET, ROBO1, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX11, SOX17, TBC1D1, TBX18, TFAP2A, TNXB, TRAP1, UMOD, UPK3A, WBP11, WNT4 Renale Hypodysplasie/Aplasie und Agenesie: 25 Gene (75,9 kb) ANOS1, BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GFRA1, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4 Vesikoureteraler Reflux (VUR): 10 Gene (33,7 kb) DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB Branchiootorenales Syndrom (BOR): 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A Renale tubuläre Dysgenesie (RTD): 4 Gene (7,7 kb) ACE, AGT, AGTR1, REN Fraser-Syndrom (FRARS): 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1 MMIH-Syndrom (MMIHS): 5 Gene (15,1 kb) ACTG2, LMOD1, MYH11, MYL9, MYLK	4 - 6 Wo	E
Meckel-Syndrom (MKS) * Gen-Panel: ID032.02, 13 Gene (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 Wo	E
Metabolische Nierenerkrankungen * Gen-Panel: ID705.00, 29 Gene (53,4 kb) AGXT, APOA1, ATP7B, B2M, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, FAH, FGA, GALT, GLA, GRHPR, GSN, HOGA1, KCNJ1, LYZ, MEFV, MVK, NLRC4, NLRP12, NLRP3, OCRL, PLCG2, SLC12A1, SLC26A1, TNFRSF1A, TTR	3 - 5 Wo	E
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
Nephronophthise (NPHP) * Gen-Panel: ID030.02, 22 Gene (70,9 kb) ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	3 - 5 Wo	E
Nierenzellkarzinom * Gen-Panel: ID041.04, 34 Gene (76,8 kb) BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1	3 - 5 Wo	E
Polyzystische Lebererkrankung (PCLD) * Gen-Panel: ID305.01 Polyzystische Lebererkrankung (PCLD): 11 Gene (45,6 kb) ALG5, ALG8, DNAJB11, DZIP1L, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63 Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 4 Gene (10,3 kb) ALG8, LRP5, PRKCSH, SEC63 Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 7 Gene (35,3 kb) ALG5, DNAJB11, DZYP1L, GANAB, PKD1, PKD2, PKHD1	3 - 5 Wo	E
Polyzystische Nierenerkrankung (PKD) * Gen-Panel: ID295.01 Polyzystische Nierenerkrankung (PKD): 7 Gene (35,3 kb) ALG5, DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1 Polyzystische Nierenerkrankung, autosomal-dominant (PKD, ADPKD): 5 Gene (20,7 kb) ALG5, DNAJB11, GANAB, PKD1, PKD2 Polyzystische Nierenerkrankung, autosomal-rezessiv (PKD, ARPKD): 2 Gene (14,5 kb) DZIP1L, PKHD1	3 - 5 Wo	E
Primäre Aminoazidurie * Gen-Panel: ID318.00 Primäre 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

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) *	3 - 5 Wo	E
Gen-Panel: ID250.01		
Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PAH): 8 Gene (23,8 kb)		
CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4		
Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb)		
NR3C2, SCNN1A, SCNN1B, SCNN1G		
Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb)		
CUL3, KLHL3, WNK1, WNK4		
Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb)		
SCNN1A, SCNN1B, SCNN1G		
Renale Amyloidose *	3 - 5 Wo	E
Gen-Panel: ID320.00, 8 Gene (11,8 kb)		
APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TTR		
Renale Hypodysplasie, Aplasie und Agenesie *	3 - 5 Wo	E
Gen-Panel: ID319.00, 23 Gene (72,5 kb)		
BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GREB1L, HNF1B, ITGA8*, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4		
Renale tubuläre Azidose (RTA) *	3 - 5 Wo	E
Gen-Panel: ID297.00, 9 Gene (18,5 kb)		
ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72		
Renale tubuläre Dysgenesie (RTD) *	2 - 4 Wo	E
Gen-Panel: ID316.00, 4 Gene (7,7 kb)		
ACE, AGT, AGTR1, REN		
Renotubuläres Fanconi-Syndrom (FRTS) *	3 - 5 Wo	E
Gen-Panel: ID359.00, 7 Gene (10,4 kb)		
CTNS, EHHADH, GATM, HNF4A, NDUFAF6, SLC2A2, SLC34A1		
Senior-Loken-Syndrom (SLSN) *	3 - 5 Wo	E
Gen-Panel: ID029.01, 8 Gene (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Thrombotische Mikroangiopathie (TMA) *	3 - 5 Wo	E
Gen-Panel: ID707.00		
Thrombotische Mikroangiopathie (TMA): 23 Gene (44,7 kb)		
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD		
Thrombotisch-thrombozytopenische Purpura (TTP): 1 Gen (4,3 kb)		
ADAMST13		
Atypisches hämolytisch-urämisches Syndrom (AHUS): 23 Gene (44,7 kb)		
ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PLG, THBD		
Tubulointerstitielle Nierenerkrankung, autosomal-dominant (ADTKD) *	3 - 5 Wo	E
Gen-Panel: ID296.00, 6 Gene (8,8 kb)		
DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD		
Urolithiasis, Nephrolithiasis und Nephrokalzinose *	3 - 5 Wo	E
Gen-Panel ID231.04		
Urolithiasis, Nephrolithiasis und Nephrokalzinose: 37 Gene (68,8 kb)		
ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, BSND, CASR, CLCN5, CLDN16, CLDN19, CLCNKB, CYP24A1, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, OCRL, OXGR1, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, XDH		
Hyperkalziurie: 15 Gene (30,2 kb)		
ADCY10, BSND, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, RRAGD, SLC12A1, SLC34A1, SLC34A3		
Hyperoxalurie: 5 Gene (6,3 kb)		
AGXT, GRHPR, HOGA1, SLC26A1, OXGR1		
Hyperglycinurie: 3 Gene (5,2 kb)		
SLC6A19, SLC6A20, SLC36A2		
Hypocitraturie: 4 Gene (11,1 kb)		
TP6V0A4, ATP6V1B1, SLC4A1, SLC4A4		
Xanthinurie: 2 Gene (6,7 kb)		
MOCOS, XDH		
Cystinurie: 2 Gene (3,5 kb)		
SLC3A1, SLC7A9		
Urothelkarzinom *	3 - 5 Wo	E
Gen-Panel: ID337.00, 34 Gene (105,0 kb)		
APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC		
Vesikoureteraler Reflux (VUR) *	3 - 5 Wo	E
Gen-Panel: ID314.00, 10 Gene (33,7 kb)		
DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB		
Wilms-Tumor (WT) *	3 - 5 Wo	E
Gen-Panel: ID335.00, 21 Gene (70,4 kb)		
AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, GPC3, GPC4, NSD1, PALB2, POU6F2, REST, TRIM28, TRIM37, TRIP13, WT1		

Erkrankung/Diagnostik	Dauer	Material
Nierenerkrankungen		
Zystische Nierenerkrankungen, umfassende Diagnostik * Gen-Panel: ID100.07 Zystische Nierenerkrankungen, umfassende Diagnostik: 49 Gene (159,8 kb) ALG5, ALG8, ALG9, ANKS6, BICC1, CEP83, CEP164, CEP290, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, REN, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, SLC41A1, TSC1, TSC2, TTC21B, TMEM67, UMOD, VHL, WDR19, XPNPEP3, ZNF423 Polyzystische Nierenerkrankung (PKD): 7 Gene (35,3 kb) ALG5, DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1 Polyzystische Lebererkrankung mit Nierenzysten (PCLD): 4 Gene (10,3 kb) ALG8, LRP5, PRKCSH, SEC63 Medulläre zystische Nierenerkrankung (MCKD, ADTKD): 5 Gene (7,7 kb) HNF1B, MUC1, REN, SEC61A1, UMOD Nephronophthise (NPHP): 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 Tuberöse Sklerose (TSC): 2 Gene (8,9 kb) TSC1, TSC2	4 - 6 Wo	E
Skelett- und Knochenerkrankungen		
Adams-Oliver-Syndrom (AOS) * Gen-Panel: ID259.00, 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 Wo	E
Akrozephalosyndaktylie (ACS) * Gen-Panel: ID311.00, 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1	3 - 5 Wo	E
Arachnodaktylie * Gen-Panel: ID124.00, 13 Gene (35,9 kb) CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFBR1, TGFBR2, SMAD3	3 - 5 Wo	E
Arthrogrypose * Gen-Panel: ID200.01 Arthrogrypose: 61 Gene (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNM2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2 Arthrogryposis multiplex congenita (AMC): 6 Gene (58,2 kb) ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1 Distale Arthrogrypose (DA): 11 Gene (40,3 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1 Letales kongenitales Kontraktursyndrom (LCCS): 11 Gene (31,5 kb) ADCY6, ADGRG6, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42 Fetale Akinesie-Deformation-Sequenz (FADS): 4 Gene (7,6 kb) DOK7, MUSK, NUP88, RAPSN	4 - 6 Wo	E
Brachydaktylie (BD) * Gen-Panel: ID218.02 Brachydaktylie (BD): 21 Gene (60,1 kb) ADAMTS10, ADAMTS17, BMP2, BMPR1B, CHST11, CHSY1, FBN1, GDF5, HOXD13, HUWE1, IHH, LTBP2, NOG, PDE3A, PITX1, PRMT7, PTHLH, ROR2, RUNX2, TBC1D24, TRPV4 Brachydaktylie (BD), nicht-syndromal: 8 Gene (10,5 kb) BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PTHLH, ROR2 Brachydaktylie (BD), syndromal: 16 Gene (52,5 kb) ADAMTS10, ADAMTS17, CHST11, CHSY1, FBN1, GDF5, HOXD13, HUWE1, LTBP2, NOG, PDE3A, PITX1, PRMT7, RUNX2, TBC1D24, TRPV4	3 - 5 Wo	E
Distale Arthrogrypose (DA) * Gen-Panel: ID196.02, 11 Gene (40,5 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1	3 - 5 Wo	E
Fetale Akinesie-Sequenz (FADS) * Gen-Panel: ID201.00, 10 Gene (30,3 kb) CHRNA1, CHRND, CHRNG, DOK7, GBE1, MUSK, MYOD1, NUP88, RAPSN, RYR1	3 - 5 Wo	E
Fraser-Syndrom (FRASRS) * Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
Frontonasale Dysplasie (FND) * Gen-Panel ID339.00: 11 Gene (22,1 kb) ALX1, ALX3, ALX4, ANKH, EFNB1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6	3 - 5 Wo	E
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

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

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Lippen-, Kiefer- und Gaumenspalte (OFC) *	4 - 6 Wo	E
Gen-Panel: ID266.00		
Lippen-, Kiefer- und Gaumenspalte (OFC): 41 Gene (129,7 kb)		
ARHGAP29, BMP4, C2CD3, CDH1, CHD7, CPLANE1, CTNND1, DDX59, DHCR7, DLX4, ESCO2, FGFR1, FGFR2, FLNB, GRHL3, HDAC8, IFT57, INTU, IRF6, KDM6A, KIAA0753, KMT2D, MEIS2, MID1, MSX1, NECTIN1, NIPBL, OFD1, RAD21, RIPK4, SEMA3E, SLC26A2, SMC1A, SMC3, SPECC1L, SUMO1, TBX22, TCTN3, TGDS, TMEM107, TP63		
Orofaziale Spalte (OFC), nicht-syndromal: 10 Gene (15,4 kb)		
ARHGAP29, BMP4, DLX4, GRHL3, IRF6, MSX1, NECTIN1, SUMO1, TBX22, TP63		
Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Cornelia-de-Lange-Syndrom (CDLS): 5 Gene (18,8 kb)		
HDAC8, NIPBL, RAD21, SMC1A, SMC3		
Kabuki-Syndrom (KABUK): 2 Gene (20,8 kb)		
KDM6A, KMT2D		
CHARGE-Syndrom: 3 Gene (12,9 kb)		
CHD7, SEMA3E, TBX22		
Multiple epiphysäre Dysplasie (EDM) *	3 - 5 Wo	E
Gen-Panel: ID202.02, 11 Gene (27,7 kb)		
CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CSGALNACT1, EIF2AK3, KIF7, MATN3, SLC26A2		
Multiples Pterygium-Syndrom *	3 - 5 Wo	E
Gen-Panel: ID158.01, 8 Gene (16,8 kb)		
CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4		
Orofaziodigitales Syndrom (OFD) *	3 - 5 Wo	E
Gen-Panel: ID265.00, 9 Gene (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Osteogenesis imperfecta (OI) *	3 - 5 Wo	E
Gen-Panel: ID066.02, 21 Gene (39,2 kb)		
ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1		
Osteopetrose (OPT) und ähnliche Knochenerkrankungen *	3 - 5 Wo	E
Gen-Panel ID346.01		
Osteopetrose (OPT) und ähnliche Knochenerkrankungen: 32 Gene (68,1 kb)		
AMER1, ANKH, CA2, CLCN7, CSF1R, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, LRP6, LRRK1, OSTM1, PLEKHM1, PTDSS1, PTH1R, SLC4A2, SLC29A3, SLCO2A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP		
Osteopetrose (OPTA, OPTB): 13 Gene (30,2 kb)		
CA2, CLCN7, FERMT3, LRP5, LRP6, OSTM1, PLEKHM1, SLC4A2, SLC29A3, SNX10, TCIRG1, TNFRSF11A, TNFSF11		
Syndromale Skelettdysplasie mit erhöhter Knochendichte: 19 Gene (38,0 kb)		
AMER1, ANKH, CSF1R, CTSK, DLX3, FAM20C, GJA1, HPGD, LEMD3, LRP4, LRRK1, PTDSS1, PTH1R, SLCO2A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP		
Osteoporose *	3 - 5 Wo	E
Gen-Panel: ID115.01, 13 Gene (29,4 kb)		
CALCR, COL1A1, COL1A2, ESR1, LRG4, LRP5, PLS3, SGMS2, SLC34A1, SLC9A3R1, UGT2B17, VDR, WNT1		
Pierre-Robin-Sequenz *	3 - 5 Wo	E
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		
Polydaktylie, nicht-syndromale Form *	3 - 5 Wo	E
Gen-Panel: ID166.02, 9 Gene (20,9 kb)		
CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141		
Rubinstein-Taybi-Syndrom (RSTS) *	3 - 5 Wo	E
Gen-Panel: ID142.01, 3 Gene (24,3 kb)		
CREBBP, EP300, SRCAP		
Skelettdysplasie, schwere Form *	4 - 6 Wo	E
Gen-Panel: ID056.01		
Skelettdysplasie, schwere Form: 46 Gene (144,2 kb)		
AGPS, ALPL, BMPER, CANT1, CEP120, CILK1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DLL3, DYNC2H1, EBP, FAM20C, FAM111A, FGFR2, FGFR3, FLNA, FLNB, GDF5, GNPAT, GPX4, HSPG2, IFT52, IFT80, IFT81, INPPL1, KIAA0586, LBR, LIFR, NEK1, NSDHL, P3H1, PEX5, PEX7, PPIB, PTH1R, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4, WDR34, WDR35		
Achondrogenesie (ACG): 4 Gene (14,1 kb)		
COL2A1, GDF5, SLC26A2, TRIP11		
Fibrochondrogenesie (FBCG): 2 Gene (10,6 kb)		
COL11A1, COL11A2		
Thanatophore Dysplasie (TD): 2 Gene (6,9 kb)		
COL1A2, FGFR3		
Chondrodysplasie, letal: 12 Gene (24,5 kb)		
AGPS, EBP, FLNB, GDF5, GNPAT, GPX4, LBR, PEX5, PEX7, PTH1R, SLC26A2, SLC35D1		
Osteogenesis imperfecta (OI), letal: 5 Gene (12,6 kb)		
CRTAP, COL1A1, COL1A2, P3H1, PPIB		
Kurzrippen-Thoraxdysplasie (SRTD), letal: 9 Gene (35,0 kb)		
CEP120, DYNC2H1, IFT52, IFT80, IFT81, KIAA0586, NEK1, WDR34, WDR35		

Erkrankung/Diagnostik	Dauer	Material
Skelett- und Knochenerkrankungen		
Skelettdysplasien, umfassende Diagnostik * Gen-Panel: ID356.00, 407 Gene (969,6 kb) ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF3, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARL6, ARSB, ARSL, ASXL1, ASXL2, ATP6V0A2, ATP7A, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRTAP, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GJA1, GLB1, GLI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GSC, GUSB, GZF1, HDAC8, HES7, HGSNAT, HHAT, HOXD13, HPGD, HS2ST1, HSPG2, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IL11RA, IL1RN, INPPL1, KAT6B, KDELR2, KIAA0753, KIF22, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MBTPS1, MEGF8, MEOX1, MESD, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NSD1, NSDHL, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RINT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMAD6, SMARCAL1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFBR2, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UFSP2, UNC45A, VDR, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6	4 - 6 Wo	E
Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD) * Gen-Panel: ID110.01 Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD): 39 Gene (90,7 kb) ACAN, ACP5, AIFM1, B3GALT6, BGN, CFAP410, CHST3, COL2A1, COL10A1, COL11A2, COMP, DDR2, DDRGK1, EXOC6B, FN1, GPX4, KIF22, MATN3, MBTPS1, MMP13, NANS, NEPRO, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, PLCB3, POP1, RPL13, RSPRY1, SIK3, SMARCAL1, TONSL, TRAPPC2, TRIP11, TRPV4, UFSP2, WISP3 Spondyloepiphysäre Dysplasie (SED): 9 Gene (25,9 kb) ACAN, CHST3, COL2A1, COMP, MBTPS1, SMARCAL1, TRAPPC2, TRPV4, WISP3 Spondylometaphysäre Dysplasie (SMD): 11 Gene (30,2 kb) ACP5, CFAP410, COL2A1, COL10A1, FN1, GPX4, PAM16, PCYT1A, PLCB3, TRIP11, TRPV4 Spondyloepimetaphysäre Dysplasie (SEMD): 21 Gene (47,6 kb) ACAN, AIFM1, B3GALT6, BGN, COL2A1, DDR2, DDRGK1, EXOC6B, KIF22, MATN3, MMP13, NANS, NEPRO, PAPSS2, PISD, POP1, RPL13, RSPRY1, SIK3, TONSL, UFSP2	3 - 5 Wo	E
Spondylokostale Dysostose (SCDO) * Gen-Panel: ID227.00, 7 Gene (14,4 kb) DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6	3 - 5 Wo	E
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		
Adipositas * Gen-Panel: ID183.02 Adipositas: 54 Gene (130,1 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FFAR4, FTO, IFT27, IFT74, IFT172, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 19 Gene (27,1 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FFAR4, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 36 Gene (103,6 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, IFT172, IFT27, IFT74, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, 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
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
Folatstoffwechselstörung * Gen-Panel: ID334.00, 10 Gene (18,7 kb) CBS*, FOLR1, FOLR2, FTCD, MTHFD1, MTHFR, MTR, MTRR, SLC19A1, SLC46A1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Glykogenspeicherkrankheit (GSD) *	3 - 5 Wo	E
Gen-Panel: ID108.00, 23 Gene (46,9 kb)		
AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4		
Glykosylphosphatidylinositol-Biosynthesedefekt (GPIBD) *	3 - 5 Wo	E
Gen-Panel: ID291.00		
Glykosylphosphatidylinositol-Biosynthesedefekt (GPIBD): 22 Gene (33,3 kb)		
GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY		
Multiple kongenitale Anomalien-Hypotonie-Krampfanfälle-Syndrom (MCAHS): 4 Gene (7,7 kb)		
PIGA, PIGN, PIGQ, PIGT		
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS): 6 Gene (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		
Hämochromatose (HFE) und Hämosiderose *	3 - 5 Wo	E
Gen-Panel: ID114.04, 10 Gene (14,6 kb)		
BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2		
Homocystinurie *	3 - 5 Wo	E
Gen-Panel: ID191.01, 9 Gene (15,3 kb)		
ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1		
Hyperinsulinämische Hypoglykämie (HHF) *	3 - 5 Wo	E
Gen-Panel: ID126.00, 8 Gene (16,9 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A		
Hyperkalzämie *	3 - 5 Wo	E
Gen-Panel: ID262.00, 8 Gene (14,6 kb)		
AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1		
Hyperoxalurie	3 - 5 Wo	E
Gen-Panel: ID363.00		
Hyperoxalurie: 6 Gene (8,6 kb)		
AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6		
Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb)		
AGXT, GRHPR, HOGA1		
Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb)		
OXGR1, SLC26A1, SLC26A6		
Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *	3 - 5 Wo	E
Gen-Panel: ID292.00, 6 Gene (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung *	3 - 5 Wo	E
Gen-Panel: ID280.00		
Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung: 44 Gene (85,9 kb)		
ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4		
Glykogenspeicherkrankheit (GSD): 24 Gene (48,4 kb)		
AGL, ALDOA. ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4		
Hyperinsulinämische Hypoglykämie (HHF): 9 Gene (18,8 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A		
Hypomagnesiämie (HOMG) *	3 - 5 Wo	E
Gen-Panel ID054.02: 14 Gene (30,9 kb)		
ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6		
Hypophosphatasie, Hypophosphatämie und Rachitis *	3 - 5 Wo	E
Gen-Panel: ID269.03		
Hypophosphatasie, Hypophosphatämie und Rachitis: 16 Gene (27,8 kb)		
ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR		
Hypophosphatämische Rachitis (HR): 9 Gene (16,1 kb)		
CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3		
Vitamin-D-abhängige hypophosphatämische Rachitis (VDDR): 4 Gene (5,8 kb)		
CYP2R1, CYP3A4, CYP27B1, VDR		
Hypophosphatasie (HPP): 1 Gen (1,6 kb)		
ALPL		
Kongenitale Störung der Glykosylierung (CDG) *	3 - 5 Wo	E
Gen-Panel ID035.02		
Kongenitale Störung der Glykosylierung (CDG): 51 Gene (74,7 kb)		
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3		
Kongenitale Störung der Glykosylierung, Typ I (CDG1): 29 Gene (39,6 kb)		
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3		
Kongenitale Störung der Glykosylierung, Typ II (CDG2): 22 Gene (35,1 kb)		
ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199		

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Kombinierter Defekt der oxidativen Phosphorylierung (COXPD) *	3 - 5 Wo	E
Gen-Panel: ID287.00, 51 Gene (67,7 kb)		
AARS2, AIFM1, ATP5F1A, C1QBP, CARS2, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL44, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NSUN3, QRSL1, PNPT1, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2		
Metabolische Epilepsien *	4 - 6 Wo	E
Gen-Panel: ID303.01		
Metabolische Epilepsien: 84 Gene (122,3 kb)		
ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1		
Glycin-Enzephalopathie (GCE): 5 Gene (8,0 kb)		
AMT, GCSH, GLDC, LIAS, SLC6A9		
Molybdän-Cofaktor-Defizienz (MOCOD): 3 Gene (3,7 kb)		
GPHN, MOCS1, MOCS2		
Zerebrales Kreatinmangelsyndrom (CCDS): 3 Gene (3,9 kb)		
GAMT, GATM, SLC6A8		
Ahornsirupkrankheit (MSUD): 5 Gene (6,6 kb)		
BCKDHA, BCKDHB, DBT, DLD, PPM1K		
2-Hydroxy-Glutarazidurie: 3 Gene (5,3 kb)		
L2HGDH, D2HGDH, IDH2, SLC25A1		
GM2-Gangliosidose: 3 Gene (3,8 kb)		
HEXA, HEXB, GM2A		
Neuronale Ceroid-Lipofuszinose (CLN): 12 Gene (14,3 kb)		
CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1		
MODY-Diabetes *	3 - 5 Wo	E
Gen-Panel: ID048.01, 14 Gene (22,9 kb)		
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1		
Mukopolysaccharidose (MPS) *	3 - 5 Wo	E
Gen-Panel: ID308.00, 12 Gene (21,2 kb)		
ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A		
Muskeldystrophie-Dystroglykanopathie (MDDG) *	3 - 5 Wo	E
Gen-Panel: ID179.00		
Muskeldystrophie-Dystroglykanopathie (MDDG): 15 Gene (24,0 kb)		
B3GALNT2, B4GAT1, DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1		
Muskeldystrophie-Dystroglykanopathie (MDDGA) mit Hirn- und Augenanomalien: 14 Gene (22,8 kb)		
B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1		
Muskeldystrophie-Dystroglykanopathie (MDDGB) mit oder ohne intellektuelle Entwicklungsstörung: 8 Gene (13,1 kb)		
DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2		
Muskeldystrophie-Dystroglykanopathie (MDDGC), Gliedergürtelmuskeldystrophie: 11 Gene (18,5 kb)		
DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, POMGNT1, POMGNT2, POMK, POMT1, POMT2		
Neonataler Diabetes mellitus *	3 - 5 Wo	E
Panel: ID162.01		
Neonataler Diabetes mellitus: 29 Gene (53,6 kb)		
ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57		
Permanenter neonataler Diabetes mellitus (PNDM): 10 Gene (15,9 kb)		
ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57		
Syndromaler neonataler Diabetes mellitus: 21 Gene (40,5 kb)		
BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5		
Neuronale Ceroid-Lipofuszinose (CLN) *	3 - 5 Wo	E
Gen-Panel: ID132.01, 15 Gene (20,2 kb)		
ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1		
Porphyrie *	3 - 5 Wo	E
Gen-Panel: ID153.01, 10 Gene (12,8 kb)		
ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS		
Primäre Aminoazidurie *	3 - 5 Wo	E
Gen-Panel: ID318.00		
Primäre 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		
Pulmonale Surfactant-Stoffwechselstörung (SMDP) *	3 - 5 Wo	E
Gen-Panel: ID168.01, 6 Gene (12,0 kb)		
ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB		

Erkrankung/Diagnostik	Dauer	Material
Stoffwechselerkrankungen		
Renale Amyloidose *	3 - 5 Wo	E
Gen-Panel: ID320.00, 8 Gene (11,8 kb)		
APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TTR		
Speicherkrankheiten mit Herzbeteiligung *	3 - 5 Wo	E
Gen-Panel: ID149.01, 13 Gene (21,5 kb)		
ATP7B, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, PRKAG2, SLC40A1, TFR2, TTR		
Störung der Peroxisomenbiogenese (PBD) *	3 - 5 Wo	E
Gen-Panel: ID083.01		
Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
Neonatale Adrenoleukodystrophie / Infantiles Refsum-Syndrom (PBD, Typ B): 11 Gene (17,0 kb)		
PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26		
Heimler-Syndrom (PBD, Typ C): 2 Gene (6,8 kb)		
PEX1, PEX6		
Stoffwechselstörung mit Epilepsie im Neugeborenenalter *	3 - 5 Wo	E
Gen-Panel: ID135.00, 25 Gene (39,4 kb)		
ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO		
Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter *	3 - 5 Wo	E
Gen-Panel: ID171.00, 18 Gene (31,5 kb)		
ALDH5A1, ATP7A, BTD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1		
Stoffwechselstörung mit Epilepsie im Schul- und Jugendalter *	3 - 5 Wo	E
Gen-Panel: ID172.00, 15 Gene (28,1 kb)		
ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2		
Zellweger-Syndrom (ZWS) *	3 - 5 Wo	E
Gen-Panel: ID084.00, 14 Gene (22,3 kb)		
ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
3-Methylglutaconazidurie (MGCA) *	3 - 5 Wo	E
Gen-Panel: ID249.01, 16 Gene (18,9 kb)		
AGK, ATPAF2, ATP5F1D, ATP5F1E, AUH, CLPB, DNAJC19, ECHS1, HTRA2, OPA3, POLG, SERAC1, SUCLA2, TAFAZZIN, TIMM50, TMEM70		
Tumorerkrankungen		
BRCA1- und BRCA2-assoziierte Tumordisposition *, #	2 - 4 Wo	E
Gen-Panel: ID001.00, 2 Gene (15,8 kb)		
BRCA1, BRCA2		
Chromosomen-Instabilitätssyndrome *	4 - 6 Wo	E
Gen-Panel: ID326.01, 40 Gene (121,0 kb)		
ANAPC1, ATM, BLM, BRCA1, BRCA2, BRIP1, DDB2, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, MRE11, NBN, PALB2, PCNA, POLH, RAD50, RAD51, RAD51C, RECQL4, RFWD3, SLX4, TOP3A, UBE2T, WRN, XPA, XPC, XRCC2		
Cowden-Syndrom (CWS) *	3 - 5 Wo	E
Gen-Panel: ID075.01, 8 Gene (12,8 kb)		
AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1		
DNA-Reparatur-Defizienz-Syndrome, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID348.00, 221 Gene (507,3 kb)		
ABRAXAS1, ALKBH2, ALKBH3, ANAPC1, APEX1, APEX2, APLF, APTX, ATM, ATR, ATRIP, ATRX, BARD1, BLM, BRCA1, BRCA2, BRIP1, CCNH, CDK7, CETN2, CHAF1A, CHEK1, CHEK2, CLK2, DCLRE1A, DCLRE1B, DCLRE1, DDB1, DDB2, DMC1, DNA2, DNPH1, DNTT, DUT, EME1, EME2, ENDOV, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, EXO1, EXO5, FAAP100, FAAP20, FAAP24, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FEN1, GEN1, GTF2E2, GTF2H1, GTF2H2, GTF2H3, GTF2H4, GTF2H5, H2AX, HELQ, HERC2, HFM1, HLTF, HMCES, HUS1, LIG1, LIG3, LIG4, MAD2L2, MBD4, MDC1, MGMT, MLH1, MLH3, MMS19, MNAT1, MPG, MPLKIP, MRE11, MSH2, MSH3, MSH4, MSH5, MSH6, MUS81, MUTYH, NABP2, NBN, NEIL1, NEIL2, NEIL3, NHEJ1, NTHL1, NUDT1, NUDT15, NUDT18, OGG1, PALB2, PARG, PARK7, PARP1, PARP2, PARP3, PARPBP, PAXIP1, PCNA, PDS5B, PER1, PMS1, PMS2, PNKP, POLA1, POLB, POLD1, POLD2, POLD3, POLD4, POLE, POLE2, POLE3, POLE4, POLG, POLH, POLI, POLK, POLL, POLM, POLN, POLQ, PRIMPOL, PRKDC, PRPF19, RAD1, RAD17, RAD18, RAD23A, RAD23B, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54B, RAD54L, RAD9A, RBBP8, RDM1, RECQL, RECQL4, RECQL5, REV1, REV3L, RIF1, RFWD3, RMI1, RNF168, RNF4, RNF8, RPA1, RPA2, RPA3, RPA4, RRM2B, SEM1, SETMAR, SHLD1, SHLD2, SHLD3, SHPRH, SLX1A, SLX1B, SLX4, SMC5, SMC6, SMUG1, SPIDR, SPO11, SPRTN, SWI5, SWSAP1, TDG, TDP1, TDP2, TOP3A, TOPBP1, TP53, TP53BP1, TREX1, TREX2, UBE2A, UBE2B, UBE2N, UBE2T, UBE2V2, UNG, USP1, UVSSA, WDR48, WRN, XAB2, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, XRCC5, XRCC6, ZSWIM7		
Dyskeratosis congenita (DKC) *	3 - 5 Wo	E
Gen-Panel: ID347.01, 15 Gene (24,1 kb)		
ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53		
Endometriumkarzinom *	3 - 5 Wo	E
Gen-Panel: ID364.00, 12 Gene (29,1 kb)		
EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, STK11, TP53		
Endometriumkarzinom, umfassende Diagnostik *	3 - 5 Wo	E
Gen-Panel: ID365.00, 26 Gene (92,9 kb)		
ATM, APC, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11, TP53		

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Erbliche Tumorerkrankungen, umfassende Diagnostik * Gen-Panel: ID018.03, 190 Gene (454,1 kb) ABRAXAS1, ACD, AIP, AKT1, ALK, ANAPC1, ANKRD26, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CCND1, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, EFL1, EGFR, ELAC2, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, FOXE1, GALNT12, GATA2, GPC3, GREM1, HABP2, HAVCR2, HNF1A, HNF1B, HOXB13, HRAS, IKZF1, KIF1B, KIT, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MAX, MC1R, MEN1, MET, MINPP1, MITF, MLH1, MLH3, MRAS, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NDUFA13, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, NTHL1, PALB2, PALLD, PARN, PAX5, PCNA, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RABL3, RAD50, RAD51, RAD51C, RAD51D, RAF1, RASA2, RB1, RECQL, RECQL4, REST, RET, RFWD3, RHBDF2, RINT1, RIT1, RNASEL, RNF139, RNF43, RPS20, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SOS2, SPRED1, SRGAP1, STK11, SUFU, TERT, TINF2, TMEM127, TOP3A, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2, XRCC3 Krebserkrankungen im Kindesalter: 128 Gene (317,3 kb) ACD, ALK, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, EFL1, ELP1, EPCAM, 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, MAP2K1, MAP2K2, MAPK1, MRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PMS2, POLH, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RAD51, RAD51C, RAF1, RB1, RECQL4, REST, RET, 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 Krebserkrankungen im Erwachsenenalter: 131 Gene (309,8 kb) AKT1, APC, ATM, BAP1, BARD1, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, CBL, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, DDB2, DICER1, DLST, EGFR, ELAC2, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, FLCN, FOXE1, GALNT12, GPC3, GREM1, HABP2, HNF1A, HNF1B, HOXB13, HRAS, KIF1B, KIT, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MAX, MC1R, MEN1, MET, MINPP1, MITF, MLH1, MLH3, MRAS, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NKX2-1, NRAS, NTHL1, PALB2, PDGFRA, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PTCH1, PTCH2, PTEN, PTPN11, RABL3, RAD51, RAD51C, RAD51D, RAF1, RB1, RET, RFWD3, RHBDF2, RIT1, RNASEL, RNF139, RNF43, RRAS2, RTEL1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, SRGAP1, STK11, SUFU, TERT, TMEM127, TP53, TSC1, TSC2, UBE2T, VHL, WRAP53, WT1, XPA, XPC, XRCC2, XRCC3	4 - 6 Wo	E
Fanconi-Anämie (FANC) * Gen-Panel: ID043.02, 21 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E
Gastrointestinaler Stromatumor (GIST) * Gen-Panel: ID226.00, 8 Gene (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 Wo	E
Glioblastom (GLM) * Gen-Panel: ID313.00, 16 Gene (61,0 kb) APC, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POT1, PTEN, TP53, TSC1, TSC2	3 - 5 Wo	E
Gorlin-Goltz-Syndrom (BCNS) * Gen-Panel: ID174.00, 3 Gene (9,4 kb) PTCH1, PTCH2, SUFU	2 - 4 Wo	E
Knochen- und Weichteilsarkome * Gen-Panel: ID223.01, 37 Gene (107,5 kb) APC, ATM, ATR, BLM, CDKN1C, CDKN2A, CHEK2, DICER1, EPCAM, ERCC2, EXT1, EXT2, FH, HRAS, KIT, MLH1, MSH2, MSH6, MTAP, NBN, NF1, PDGFRA, PMS2, PRKAR1A, PTCH1, RB1, RECQL4, SDHA, SDHB, SDHC, SDHD, SQSTM1, SUFU, TNFRSF11A, TP53, WRN, ZNF687	4 - 6 Wo	E
Kolorektales Karzinom (CRC) * Gen-Panel: ID049.01, 14 Gene (40,4 kb) ATM, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, 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
Kolorektales Karzinom, umfassende Diagnostik * Gen-Panel: ID285.01, 33 Gene (87,9 kb) APC, ATM, AXIN2, BLM, BMPR1A, CDH1, CHEK2, EPCAM, EXO1, FLCN, GALNT12, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PLA2G2A, PMS2, POLD1, POLE, PTEN, RFC1, RPA1, RNF43, RPS20, SMAD4, STK11, TGFBR2, TP53	3 - 5 Wo	E
Kolorektales Karzinom und Polyposis * Gen-Panel ID006.08 Kolorektales Karzinom und Polyposis: 22 Gene (62,3 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Kolorektale Polyposis: 14 Gene (37,6 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11 Kolorektales Karzinom: 14 Gene (40,4 kb) ATM, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11	3 - 5 Wo	E
Konstitutionelles MMR-Defizienz-Syndrom (CMMRDS, MMRCS) * Gen-Panel: ID362.00, 5 Gene (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	2 - 4 Wo	E
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

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Krebserkrankungen im Kindesalter * Gen-Panel: ID333.00 Krebserkrankungen im Kindesalter: 139 Gene (341,1kb) ACD, ALK, ANKRD26, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNAJC21, EFL1, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, GATA2, GPC3, HRAS, IKZF1, KIF1B, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PMS2, POLE, POLH, POU6F2, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RAD51, RAD51C, RAF1, RB1, RECQL4, REST, RET, RFWD3, RIT1, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, SRP72, STK11, SUFU, TERT, TMEM127, TINF2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2 Maligne hämatologische Erkrankungen: 49 Gene (126,4 kb) ACD, ANKRD26, ATM, BLM, BRCA1, BRCA2, BRIP1, CEBPA, DDX41, DKC1, DNAJC21, EFL1, ELANE, ERCC4, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA2, IKZF1, MAD2L2, NBN, NHP2, NOP10, PALB2, PARN, PAX5, RAD51, RAD51C, RFWD3, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SRP72, TERT, TINF2, TP53, UBE2T, WRAP53, XRCC2 Tumoren des Zentralnervensystems: 32 Gene (111,9 kb) ALK, APC, BRCA2, CDKN2A, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, KIF1B, LZTR1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PALB2, PHOX2B, PMS2, PTCH1, PTCH2, PTEN, RB1, SMARCA4, SMARCB1, SUFU, TP53, TSC1, TSC2, VHL Endokrine Tumoren: 15 Gene (21,2 kb) CDC73, CDKN1B, DLST, KIF1B, MAX, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL RASopathien: 18 Gene (36,6 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2 Wilms-Tumor (WT): 10 Gene (29,5 kb) BRCA2, CDKN1C, GPC3, DIS3L2, POU6F2, TRIM28, WT1, TRIM37, CDC73, REST Xeroderma pigmentosum (XP): 9 Gene (19,0 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC MMR-Defizienz-Syndrom (MMRCS): 5 Gene (12,7 kb) MLH1, MSH2, PMS2, MSH6, EPCAM	4 - 6 Wo	E
Lungenkarzinom * Gen-Panel: ID260.02, 33 Gene (122,5 kb) ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTYH, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53	4 - 6 Wo	E
Lynch-Syndrom (LYNCH, HNPCC) * Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	2 - 4 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
Magenkarzinom * Gen-Panel: ID090.03, 24 Gene (71,9 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	3 - 5 Wo	E
Mammakarzinom * Gen-Panel: ID021.02, 12 Gene (41,0 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53	2 - 4 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Tumorerkrankungen		
Mamma- und Ovarialkarzinom (HBOC) *	3 - 5 Wo	E
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		
Mamma- und Ovarialkarzinom, umfassende Diagnostik *	4 - 6 Wo	E
Gen-Panel: ID068.01, 50 Gene (151,5 kb)		
ABRAXAS1, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RECQL, RECQL4, SDHB, SDHC, SDHD, SLX4, SMARCA4, STK11, TP53, XRCC2		
Medulloblastom (MDB) *	3 - 5 Wo	E
Gen-Panel: ID205.02, 22 Gene (75,7 kb)		
APC, BRCA2, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTCH1, PTCH2, PTEN, SMARCB1, SMARCA4, SUFU, TP53, VHL		
Neurofibromatose (NF) *, #	3 - 5 Wo	E
Gen-Panel: ID210.00, 3 Gene (11,6 kb)		
NF1, NF2, SPRED1		
Nierenzellkarzinom *	3 - 5 Wo	E
Gen-Panel: ID041.04, 34 Gene (76,8 kb)		
BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1		
Ovarialkarzinom *	3 - 5 Wo	E
Gen-Panel: ID004.04, 14 Gene (45,5 kb)		
BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53		
Pankreaskarzinom *	3 - 5 Wo	E
Gen-Panel: ID089.03, 17 Gene (55,2 kb)		
APC, ATM, BRCA1, BRCA2, CDKN2A, CTRC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL		
Paragangliom und Phäochromozytom *	3 - 5 Wo	E
Gen-Panel: ID042.02, 16 Gene (29,4 kb)		
DLST, FH, KIF1B, GDNF, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL		
Plasmozytom *	3 - 5 Wo	E
Gen-Panel: ID354.01, 40 Gene (89,5 kb)		
ARID1A, ATM, BLM, BTK, CASP8, CASP10, CDKN2A, CBL, CTLA4, DDX41, EFL1, ETV6, FANCA, FAS, FASLG, HCLS1, KDM1A, KLHDC8B, KRAS, LAPTM5, MLH1, MSH2, MSH6, MYD88, NBN, NF1, NRAS, PAX5, PRF1, PMS2, POT1, PRKCD, PTPN11, RBM8A, SBDS, SH2B3, SH2D1A, TP53, USP45, WAS		
Polyposis-Syndrom (PS, FAP) *	3 - 5 Wo	E
Gen-Panel: ID005.05, 14 Gene (37,6 kb)		
APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11		
Prostatakarzinom *	3 - 5 Wo	E
Gen-Panel: ID140.01, 28 Gene (91,1 kb)		
AR, ATM, ATR, BAP1, BRCA1, BRCA2, CYP3A43, CDH1, CHEK2, EHBP1, ELAC2, EPCAM, HOXB13, MLH1, MRE11A, MSR1, MSH2, MSH6, MSMB, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, RNASEL, TP53, TRRAP		
Schilddrüsenkarzinom *	3 - 5 Wo	E
Gen-Panel: ID220.01, 26 Gene (54,7 kb)		
AKT1, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, PIK3CA, PRKAR1A, PTEN, RET, SDHA, SDHB, SDHC, SDHD, SEC23B, SRGAP1, TP53		
Urothelkarzinom *	3 - 5 Wo	E
Gen-Panel: ID337.00, 34 Gene (105,0 kb)		
APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC		
Wilms-Tumor (WT) *	3 - 5 Wo	E
Gen-Panel: ID335.00, 21 Gene (70,4 kb)		
AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, GPC3, GPC4, NSD1, PALB2, POU6F2, REST, TRIM28, TRIM37, TRIP13, WT1		
Xeroderma pigmentosum (XP) *	3 - 5 Wo	E
Gen-Panel: ID282.00, 10 Gene (23,5 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC		

Erkrankung/Diagnostik	Dauer	Material
Ziliopathien		
Bardet-Biedl-Syndrom (BBS) *	3 - 5 Wo	E
Gen-Panel: ID093.02, 21 Gene (39,0 kb)		
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Joubert-Syndrom (JBTS) *	4 - 6 Wo	E
Gen-Panel: ID028.03, 40 Gene (104,1 kb)		
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGAGRAM1, TTC21B, ZNF423		
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD) *	3 - 5 Wo	E
Gen-Panel: ID067.00		
Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD): 20 Gene (68,2 kb)		
CEP120, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60		
Kurzrippen-Thoraxdysplasie (Jeune-ATD, SRPS, SRTD): 18 Gene (61,3 kb)		
CEP120, DYNC2H1, DYNC2LI1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60		
Ellis-van-Crefeld-Syndrom (EVC): 4 Gene (11,5 kb)		
DYNC2L1, EVC, EVC1, WDR35		
Meckel-Syndrom (MKS) *	3 - 5 Wo	E
Gen-Panel: ID032.02, 13 Gene (35,1 kb)		
B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231		
Nephronophthise (NPHP) *	3 - 5 Wo	E
Gen-Panel: ID030.02, 22 Gene (70,9 kb)		
ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423		
Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *	4 - 6 Wo	E
Gen-Panel: ID085.02, 42 Gene (132,9 kb)		
ARMC4, CCDC103, CCDC39, CCDC40, CCDC65, CCDC114, CCDC151, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH5, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC6, LRRC56, MCIDAS, NEK10, NME8, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC12, TTC25, ZMYND10		
Senior-Loken-Syndrom (SLSN) *	3 - 5 Wo	E
Gen-Panel: ID029.01, 8 Gene (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Präventionsdiagnostik: Carrier-Screening		
Carrier-Screening *	4 - 6 Wo	E
Gen-Panel: ID350.00, 625 Gene (1591,4 kb)		
AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACOX1, ACSF3, ADA, ADAMTS13, ADAMTS2, ADGRG1, ADGRV1, AFF2, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMT, ANK1, ANO10, ANTXR2, APTX, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRIP1, BSND, BTD, BTK, CA2, CAPN3, CASR, CBS, CC2D2A, CCDC88C, CD3D, CD3E, CD40LG, CDH23, CEP290, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNG, CIITA, CLCN1, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL1A2, COL27A1, COL4A3, COL4A4, COL4A5, COL7A1, COQ2, COQ8A, CPS1, CPT1A, CPT2, CRB1, CRLF1, CRTAP, CSTB, CTNS, CTSC, CTSD, CTSK, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP27A1, CYP27B1, D2HGDH, DBT, DCLRE1C, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DHDDS, DKC1, DLD, DLL3, DMD, DMP1, DNAH5, DNAI1, DNAI2, DNAJC19, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPYD, DSP, DUOX2, DUOXA2, DYNC2H1, DYSF, EDA, EDN3, EDNRB, EFEMP2, EGR2, EIF2AK3, EIF2B5, ELP1, EMD, ENPP1, EPB42, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, F11, F8, F9, FAH, FAM126A, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCG, FBLN5, FBP1, FGA, FGB, FGD4, FGG, FH, FKRP, FKTN, FLNA, FMO3, FOLR1, FOXN1, FOXP3, FRAS1, FREM2, FUCA1, FXN, G6PC1, G6PC3, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDF5, GFM1, GJA1, GJB1, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNE, GNPTAB, GNPTG, GNS, GP1BA, GP9, GRHPR, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HESX1, HEXA, HEXB, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HOGA1, HPD, HPRT1, HPS1, HPS3, HPS4, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPG2, HYAL1, HYLS1, IDS, IDUA, IGBP1, IGF1, IGHMBP2, IGSF1, IL2RG, IL7R, INPP5E, INS, INSR, INVS, IQCB1, ITGA6, ITGB4, IVD, IYD, JAG1, JAK3, KCNJ1, KCNJ11, KCNQ1, KCNQ2, KCTD7, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LARGE1, LBR, LCA5, LDLR, LDLRAP1, LHCGR, LHX3, LIFR, LIPA, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MED12, MED17, MEFV, MESP2, MFSD8, MGAT2, MID1, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPI, MPL, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTTP, MUTYH, MVK, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFAF5, NDUFS6, NEU1, NEUROG3, NHLRC1, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NR0B1, NR2E3, NTRK1, NUP62, OAT, OCA2, OCRL, OFD1, OPA3, OSTM1, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRT2, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRT2, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A1, SLC4A11, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMARCAL1, SMPD1, SNAP29, SP110, SPR, SRD5A2, ST3GAL5, STAR, STRA6, SUCLG1, SUMF1, SUOX, TAFAZZIN, TAT, TBCE, TCIRG1, TECPR2, TF, TFR2, TG, TGM1, TH, TIMM8A, TK2, TMEM216, TMEM67, TNFRSF11B, TNNT1, TPO, TPP1, TREX1, TRHR, TRIM32, TRIM37, TRMU, TSEN54, TSHB, TSHR, TSPYL1, TTC37, TTN, TTPA, TWNK, TYMP, TYR, TYRP1, UBA1, UBR1, UGT1A1, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WAS, WNT10A, WNT3, WNT7A, WRN, WT1, XPA, XPC, ZIC3, ZMPSTE24, ZNF469		

Erkrankung/Diagnostik	Dauer	Material
Pränatale Diagnostik: Fetale Anomalien		
Fetale Anomalien *	4 - 6 Wo	E
Gen-Panel: ID850.00, 1223 Gene (3235,6 kb)		
AAAS, ABCA12, ABCC6, ABCC9, ABHD5, ABL1, ACAD9, ACADVL, ACAN, ACE, ACOX1, ACP5, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTG2, ACVR2B, ACY1, ADAMTS10, ADAMTS17, ADAMTS3, ADAMTSL2, ADAR, ADGRG1, ADGRG6, ADNP, ADSL, AFF4, AGK, AGL, AGPS, AHCY, AHDC1, AHI1, AKT1, AKT2, AKT3, ALDH18A1, ALDH1A3, ALDH3A2, ALDH7A1, ALDOA, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALX1, ALX3, ALX4, AMACR, AMER1, AMMECR1, AMPD2, AMT, ANAPC1, ANKH, ANKRD11, ANKS6, ANOS1, ANTXR1, ANTXR2, AP1S2, AP4E1, AR, ARCN1, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6V0A2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCL11A, BCOR, BCS1L, BFSP2, BGN, BHLHA9, BICD2, BIN1, BLM, BLTP1, BMP1, BMP2, BMP4, BMPER, BMPR1B, BNC2, BPNT2, BRAF, BRAT1, BRCA2, BRIP1, BRPF1, BSND, BTD, BUB1B, C2CD3, CA2, CA8, CACNA1C, CACNA1E, CACNA1G, CANT1, CASK, CASR, CBL, CC2D2A, CCBE1, CCDC103, CCDC39, CCDC40, CCDC8, CCDC88C, CCND2, CCNQ, CDAN1, CDC45, CDC6, CDH1, CDH3, CDK13, CDK5RAP2, CDK8, CDKL5, CDKN1C, CDON, CDT1, CELSR1, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFAP53, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD4, CHD7, CHKB, CHMP1A, CHRNA1, CHRNA3, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CILK1, CIT, CKAP2L, CLCN7, CLP1, CLPB, CNOT1, CNOT3, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COLEC10, COLEC11, COLQ, COQ4, COQ9, COX7B, CPLANE1, CPT2, CRADD, CRB2, CREB3L1, CREBBP, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CSF1R, CSNK2A1, CSPP1, CTC1, CTCF, CTNNB1, CTNND1, CTSA, CTSD, CTSK, CTU2, CUL4B, CUL7, CWC27, CYP11A1, CYP11B1, CYP17A1, CYP1B1, CYP21A2, CYP26B1, CYP2U1, CYP4F22, DAG1, DARS1, DCHS1, DCX, DDR2, DDX11, DDX3X, DDX59, DENND5A, DHCR24, DHCR7, DHFR, DHODH, DIAPH1, DIS3L2, DISP1, DKC1, DLL3, DLL4, DLX5, DMPK, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB11, DNAL1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK6, DOK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSP, DSTYK, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, DYRK1A, DZIP1L, EBF3, EBP, ECEL1, EDA, EDNRA, EDNRB, EED, EFNB1, EFTUD2, EHMT1, EIF2AK3, EIF2B2, EIF2B3, EIF2S3, EIF4A3, EIF5A, ELAC2, ELN, ELOVL4, EMD, EML1, EMX2, ENPP1, EOGT, EP300, EPG5, EPHB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ESCO2, ETFA, ETFB, ETFDH, EVC, EVC2, EXOC3L2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EZH2, FAH, FAM111A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAR1, FAT4, FBLN5, FBN1, FBN2, FBXL4, FGD1, FGF10, FGF3, FGF8, FGFR1, FGFR2, FGFR3, FH, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FLNC, FLT4, FLVCR2, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMD4A, FTL, FUT8, FYCO1, FZD2, G6PC3, GAA, GALC, GALE, GALK1, GALNS, GALNT2, GANAB, GATA2, GATA3, GATA4, GATA6, GBA1, GBA2, GBE1, GCDH, GDF1, GDF5, GDF6, GFAP, GFM1, GFPT1, GJA1, GJA3, GJA8, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLI1, GLI2, GLI3, GLIS3, GLUL, GMNN, GMPPB, GNAI3, GNAO1, GNAS, GNB1, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC6, GPI, GPSM2, GREB1L, GRHL3, GRIN1, GRIN2B, GRIP1, GSC, GTF2H5, GTPBP3, GUCY2C, GUSB, GZF1, H1-4, HAAO, HADHA, HADHB, HBA1, HBA2, HCCS, HCFC1, HDAC8, HES7, HESX1, HIBCH, HIVEP2, HMGA2, HNF1B, HNF4A, HNRNPK, HOXA1, HOXA13, HOXD13, HPSE2, HR, HRAS, HSD17B3, HSD17B4, HSF4, HSPD1, HSPG2, HUWE1, HYCC1, HYLS1, IARS1, IDH1, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGF2, IGHMBP2, IHH, IKBKG, IL11RA, IL1RAPL1, INPP5E, INPPL1, INSR, INTU, INVS, IQCB1, IRF6, ITGA3, ITGA6, ITGA8, ITGB4, JAG1, KANSL1, KAT6A, KAT6B, KATNB1, KCNJ1, KCNJ2, KCTD1, KDM5C, KDM6A, KIAA0586, KIAA0753, KIF11, KIF14, KIF1A, KIF1B, KIF22, KIF2A, KIF5C, KIF7, KLF1, KLHL40, KLHL41, KLHL7, KMT2A, KMT2C, KMT2D, KNL1, KRAS, KRIT1, KYNU, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LARGE1, LARP7, LBR, LFNG, LGI4, LHX3, LHX4, LIFR, LIG4, LIPA, LMBR1, LMBRD1, LMNA, LMNB1, LMNB2, LMOD3, LMX1B, LONP1, LRP2, LRP4, LRP5, LRRC56, LTBP3, LTBP4, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAGEL2, MAP2K1, MAP2K2, MAP3K1, MAP3K20, MAP3K7, MAPRE2, MASP1, MATN3, MBTPS2, MCOLN1, MCPH1, MED12, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESD, MESP2, MFRP, MFSD2A, MGP, MID1, MKKS, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MMP13, MMP21, MN1, MNX1, MOCS1, MOCS2, MOGS, MPDU1, MPLKIP, MRAS, MRPS22, MSL3, MSMO1, MSTO1, MSX1, MSX2, MTM1, MTO1, MTOR, MTRFR, MUSK, MYBPC1, MYCN, MYH10, MYH11, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL1, MYMK, MYO18B, MYO9A, MYOCD, MYPN, MYRF, MYT1, NAA10, NACC1, NADSYN1, NAGA, NALCN, NANS, NBAS, NBN, NDE1, NDP, NDUFAF5, NEB, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEK9, NEU1, NF1, NFIX, NHEJ1, NHS, NIPAL4, NIPBL, NKX2-5, NKX3-2, NODAL, NOG, NOTCH1, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPR2, NR0B1, NR2F2, NR5A1, NRAS, NSD1, NSDHL, NUBPL, NUP107, NXN, OBSL1, OCLN, OCRL, ODAD1, ODAD2, ODAD3, OFD1, OPHN1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTX2, P3H1, P4HB, PAFAH1B1, PAK3, PALB2, PAPSS2, PARN, PAX2, PAX3, PAX6, PAX7, PAX8, PBX1, PCGF2, PCNT, PCYT1A, PDCD10, PDE4D, PDGFRB, PDHA1, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAP2, PGAP3, PGM1, PGM3, PHF6, PHF8, PHGDH, PHIP, PHOX2B, PIBF1, PIEZO1, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGT, PIGV, PIK3C2A, PIK3CA, PIK3R1, PIK3R2, PITX1, PITX2, PITX3, PKD1, PKD1L1, PKD2, PKHD1, PKLR, PLAG1, PLG, PLK4, PLOD1, PLOD2, PMM2, PNKP, PNPLA1, POC1A, POGZ, POLE, POLG2, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, 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