

# SYNLAB Humangenetik

Stand: 12/2024

## Next Generation Sequencing (NGS)

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Erkrankung/Diagnostik	Dauer	Material
<b>Augenerkrankungen</b>		
<b>Achromatopsie (ACHM) *</b> Gen-Panel: ID164.02, 6 Gene (10,4 kb) ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	3 - 5 Wo	E
<b>Albinismus, umfassende Diagnostik *</b> 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
<b>Altersbedingte Makuladegeneration (AMD, ARMD) *</b> 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
<b>Anophthalmie und Mikrophthalmie (MCOP) *</b> Gen-Panel ID263.02 Anophthalmie und Mikrophthalmie (MCOP): 46 Gene (107,6 kb) ABCB6, ALDH1A3, BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3, FZD5, GDF3, GDF6, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, MITF, NAA10, NHEJ1, OTX2, PAX2, PAX6, PITX3, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIX6, SMOG1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, TMEM98, VAX1, VSX2, YAP1 Mikrophthalmie, isolierte Form (MCOP): 6 Gene (8,6 kb) ALDH1A3, GDF3, GDF6, MFRP, PRSS56, VSX2 Mikrophthalmie, syndromale Form (MCOPS): 13 Gene (24,9 kb) BCOR, BMP4, HCCS, HMGB3, MAB21L2, NAA10, OTX2, SOX2, RARB, RAX, STRA6, TENM3, VAX1 Mikrophthalmie und Kolobom (MCOPCB): 11 Gene (22,1 kb) ABCB6, GDF3, GDF6, FZD5, NHEJ1, PAX6, RBP4, SHH, STRA6, TENM3, VSX2 Syndrome mit Mikrophthalmie: 34 Gene (90,4 kb) BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MITF, NAA10, OTX2, PAX2, PITX3, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, SIX6, SMOG1, SOX2, STRA6, TBC1D20, TFAP2A, TMEM98, TENM3, VAX1, YAP1	4 - 6 Wo	E
<b>Dysgenese des vorderen Augensegmentes (ASGD) *</b> Gen-Panel: ID182.03 Dysgenese des vorderen Augensegmentes (ASGD): 11 Gene (21,5 kb) B3GLCT, CPAMD8, CYP1B1, ELP4, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN, TRIM44 Peters-Anomalie: 7 Gene (8,8 kb) B3GLCT, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3 Axenfeld-Rieger-Anomalie: 3 Gene (3,8 kb) FOXC1, PAX6, PITX2 Aniridie (AN): 3 Gene (3,6 kb) ELP4, PAX6, TRIM44	3 - 5 Wo	E
<b>Fraser-Syndrom (FRASRS) *</b> Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
<b>Fuchs-Endotheldystrophie (FECF) *</b> Gen-Panel: ID261.00, 5 Gene (13,5 kb) AGBL1, COL8A2, SLC4A11, TCF4, ZEB1	3 - 5 Wo	E
<b>Glaukom (GLC) *</b> Gen-Panel: ID275.01 Glaukom (GLC): 27 Gene (53,5 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CPAMD8, CYP1B1, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, PAX6, PITX2, PITX3, PXDN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMC01, WDR36 Primäres Glaukom (GLC1, GLC3): 20 Gene (37,8 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CYP1B1, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMC01, WDR36 Dysgenese des vorderen Augensegmentes (ASGD): 8 Gene (17,5 kb) CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN	3 - 5 Wo	E
<b>Hermansky-Pudlak-Syndrom (HPS) *</b> Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
<b>Hohe Myopie (MYP) *</b> Gen-Panel ID079.03 Hohe Myopie (MYP): 25 Gene (81,1 kb) ARR3, CACNA1F, CNGB3, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CPSF1, GPR179, GRM6, GZF1, IRX5, LOXL3, LRPAP1, LRP2, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644 Hohe Myopie, nicht-syndromale Form (MYP): 12 Gene (24,5 kb) ARR3, CNGB3, CPSF1, LOXL3, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644 Syndrome mit hoher Myopie: 14 Gene (58,7 kb) CACNA1F, COL11A1, COL2A1, COL9A1, COL9A2, COL9A3, COL18A1, GPR179, GRM6, GZF1, LRP2, IRX5, P3H2, SLITRK6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Augenerkrankungen</b>		
<b>Hornhautdystrophie *</b> Gen-Panel: ID329.01 Hornhautdystrophie: 27 Gene (68,9 kb) AGBL1, CHST6, COL8A2, COL17A1, CYP4V2, DCN, GRHL2, GSN, KERA, KRT3, KRT12, LCAT, LOXHD1, MCOLN1, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFB1, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469 Fuchs-Endotheldystrophie (FECD): 6 Gene (20,2 kb) AGBL1, COL8A2, LOXHD1, SLC4A11, TCF4, ZEB1 Hintere polymorphe Hornhautdystrophie (PPCD): 4 Gene (8,2 kb) COL8A2, GRHL2, OVOL2, ZEB1 Gittrige Hornhautdystrophie (CDL): 3 Gene (5,4 kb) GSN, TACSTD2, TGFB1 Stromale Hornhautdystrophie: 5 Gene (11,6 kb) CHST6, DCN, PIKFYVE, TGFB1, UBIAD1 Epitheliale Hornhautdystrophie: 6 Gene (12,6 kb) COL17A1, KRT3, KRT12, MCOLN1, TACSTD2, TGFB1 Brittle-Cornea-Syndrom (BCN): 2 Gene (13,8 kb) PRDM5, ZNF468 Keratokonus (KTCN): 2 Gene (2,5 kb) TUBA3D, VSX1	3 - 5 Wo	E
<b>Katarakt (CTRCT) *</b> 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
<b>Kongenitale extraokuläre Muskelfibrose (CFEOM) *</b> Gen-Panel: ID063.00, 5 Gene (10,5 kb) COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3	3 - 5 Wo	E
<b>Kongenitaler Nystagmus (NYS) *</b> Gen-Panel ID331.01 Kongenitaler Nystagmus (NYS): 31 Gene (71,0 kb) AHR, ATF6, CACNA1F, CEP290, CNGA3, CNGB3, CRB1, CRX, DAGLA, DCT, FRMD7, GPR143, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, LRMDA, NMNAT1, NYX, OCA2, PAX6, ROBO1, RPE65, RPGRIP1, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1 Nystagmus, nicht-syndromal (NYS): 3 Gene (8,3 kb) FRMD7, GPR143, ROBO1 Foveahypoplasie, nicht-syndromal (FVH): 3 Gene (5,1 kb) AHR, PAX6, SLC38A8 Okulokutaner Albinismus (OCA): 7 Gene (11,1 kb) DCT, LRMDA, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Lebersche kongenitale Amaurose (LCA): 12 Gene (29,5 kb) CEP290, CRB1, CRX, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1	3 - 5 Wo	E
<b>Kongenitale stationäre Nachtblindheit (CSNB) *</b> 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
<b>Lebersche hereditäre Optikusneuropathie (LHON) #</b> 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
<b>Lebersche kongenitale Amaurose (LCA) *</b> 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
<b>Makuladystrophie (MD) *</b> 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
<b>Morbus Stargardt (STGD) *</b> Gen-Panel: ID102.01, 4 Gene (11,4 kb) ABCA4, ELOVL4, PROM1, PRPH2	3 - 5 Wo	E
<b>Optikusatrophie (OPA) *</b> Gen-Panel ID081.05 Optikusatrophie (OPA): 38 Gene (60,6 kb) ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, KLC2, MCAT, MECR, MFF, MFN2, MIEF1, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, RTN4IP1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, SSBP1, TBCE, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1, ZNHIT3 Optikusatrophie (OPA) nicht-syndromal: 12 Gene (18,5 kb) ACO2, AFG3L2, DNM1L, MCAT, MECR, MIEF1, OPA1, OPA3, RTN4IP1, SSBP1, TMEM126A, YME1L1 Syndrome mit Optikusatrophie: 29 Gene (46,6 kb) ATP1A3, C19ORF12, CISD2, DNAJC30, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, TBCE, TIMM8A, UCHL1, WFS1, ZNHIT3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Augenerkrankungen</b>		
<b>Okulokutaner Albinismus (OCA) *</b> Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRM1A, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E
<b>Netzhauterkrankungen, umfassende Diagnostik *</b> Gen-Panel: ID383.00, 306 Gene (764,2 kb) ABCA4, ABCO6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AFG3L2, AGLB5, AHI1, AHR, AIPL1, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARSG, ATF6, ATOH7, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP78, CERKL, CFAP20, CFAP410, CFAP418, CFH, CHM, CIB2, CLCC1, CLEC3B, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COQ2, COQ8B, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTNND1, CTSD, CWC27, CYP4V2, DCT, DHDDS, DHX38, DNAJC5, DRAM2, DYNC2H1, EFEMP1, ELOVL4, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FSCN2, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HGSNAT, HK1, HKDC1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF3B, KIF11, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MAN2B1, MAPKAPK3, MCOLN1, MED12, MERTK, MFRP, MFSDB, MIR204, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTTT, MYK, MYO7A, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, OPN1MW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDZD7, PDE6H, PDSS1, PEX1, PEX2, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, POMGNT2, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PYGM, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS2, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RRG1, RRG2, RRG3, RRG4, RRG5, RRG6, RRG7, RRG8, RRG9, RRG10, RRG11, RRG12, RRG13, RRG14, RRG15, RRG16, RRG17, RRG18, RRG19, RRG20, RRG21, RRG22, RRG23, RRG24, RRG25, RRG26, RRG27, RRG28, RRG29, RRG30, RRG31, RRG32, RRG33, RRG34, RRG35, RRG36, RRG37, RRG38, RRG39, RRG40, RRG41, RRG42, RRG43, RRG44, RRG45, RRG46, RRG47, RRG48, RRG49, RRG50, RRG51, RRG52, RRG53, RRG54, RRG55, RRG56, RRG57, RRG58, RRG59, RRG60, RRG61, RRG62, RRG63, RRG64, RRG65, RRG66, RRG67, RRG68, RRG69, RRG70, RRG71, RRG72, RRG73, RRG74, RRG75, RRG76, RRG77, RRG78, RRG79, RRG80, RRG81, RRG82, RRG83, RRG84, RRG85, RRG86, RRG87, RRG88, RRG89, RRG90, RRG91, RRG92, RRG93, RRG94, RRG95, RRG96, RRG97, RRG98, RRG99, RRG100, RRG101, RRG102, RRG103, RRG104, RRG105, RRG106, RRG107, RRG108, RRG109, RRG110, RRG111, RRG112, RRG113, RRG114, RRG115, RRG116, RRG117, RRG118, RRG119, RRG120, RRG121, RRG122, RRG123, RRG124, RRG125, RRG126, RRG127, RRG128, RRG129, RRG130, RRG131, RRG132, RRG133, RRG134, RRG135, RRG136, RRG137, RRG138, RRG139, RRG140, RRG141, RRG142, RRG143, RRG144, RRG145, RRG146, RRG147, RRG148, RRG149, RRG150, RRG151, RRG152, RRG153, RRG154, RRG155, RRG156, RRG157, RRG158, RRG159, RRG160, RRG161, RRG162, RRG163, RRG164, RRG165, RRG166, RRG167, RRG168, RRG169, RRG170, RRG171, RRG172, RRG173, RRG174, RRG175, RRG176, RRG177, RRG178, RRG179, RRG180, RRG181, RRG182, RRG183, RRG184, RRG185, RRG186, RRG187, RRG188, RRG189, RRG190, RRG191, RRG192, RRG193, RRG194, RRG195, RRG196, RRG197, RRG198, RRG199, RRG200, RRG201, RRG202, RRG203, RRG204, RRG205, RRG206, RRG207, RRG208, RRG209, RRG210, RRG211, RRG212, RRG213, RRG214, RRG215, RRG216, RRG217, RRG218, RRG219, RRG220, RRG221, RRG222, RRG223, RRG224, RRG225, RRG226, RRG227, RRG228, RRG229, RRG230, RRG231, RRG232, RRG233, RRG234, RRG235, RRG236, RRG237, RRG238, RRG239, RRG240, RRG241, RRG242, RRG243, RRG244, RRG245, RRG246, RRG247, RRG248, RRG249, RRG250, RRG251, RRG252, RRG253, RRG254, RRG255, RRG256, RRG257, RRG258, RRG259, RRG260, RRG261, RRG262, RRG263, RRG264, RRG265, RRG266, RRG267, RRG268, RRG269, RRG270, RRG271, RRG272, RRG273, RRG274, RRG275, RRG276, RRG277, RRG278, RRG279, RRG280, RRG281, RRG282, RRG283, RRG284, RRG285, RRG286, RRG287, RRG288, RRG289, RRG290, RRG291, RRG292, RRG293, RRG294, RRG295, RRG296, RRG297, RRG298, RRG299, RRG300, RRG301, RRG302, RRG303, RRG304, RRG305, RRG306, RRG307, RRG308, RRG309, RRG310, RRG311, RRG312, RRG313, RRG314, RRG315, RRG316, RRG317, RRG318, RRG319, RRG320, RRG321, RRG322, RRG323, RRG324, RRG325, RRG326, RRG327, RRG328, RRG329, RRG330, RRG331, RRG332, RRG333, RRG334, RRG335, RRG336, RRG337, RRG338, RRG339, RRG340, RRG341, RRG342, RRG343, RRG344, RRG345, RRG346, RRG347, RRG348, RRG349, RRG350, RRG351, RRG352, RRG353, RRG354, RRG355, RRG356, RRG357, RRG358, RRG359, RRG360, RRG361, RRG362, RRG363, RRG364, RRG365, RRG366, RRG367, RRG368, RRG369, RRG370, RRG371, RRG372, RRG373, RRG374, RRG375, RRG376, RRG377, RRG378, RRG379, RRG380, RRG381, RRG382, RRG383, RRG384, RRG385, RRG386, RRG387, RRG388, RRG389, RRG390, RRG391, RRG392, RRG393, RRG394, RRG395, RRG396, RRG397, RRG398, RRG399, RRG400, RRG401, RRG402, RRG403, RRG404, RRG405, RRG406, RRG407, RRG408, RRG409, RRG410, RRG411, RRG412, RRG413, RRG414, RRG415, RRG416, RRG417, RRG418, RRG419, RRG420, RRG421, RRG422, RRG423, RRG424, RRG425, RRG426, RRG427, RRG428, RRG429, RRG430, RRG431, RRG432, RRG433, RRG434, RRG435, RRG436, RRG437, RRG438, RRG439, RRG440, RRG441, RRG442, RRG443, RRG444, RRG445, RRG446, RRG447, RRG448, RRG449, RRG450, RRG451, RRG452, RRG453, RRG454, RRG455, RRG456, RRG457, RRG458, RRG459, RRG460, RRG461, RRG462, RRG463, RRG464, RRG465, RRG466, RRG467, RRG468, RRG469, RRG470, RRG471, RRG472, RRG473, RRG474, RRG475, RRG476, RRG477, RRG478, RRG479, RRG480, RRG481, RRG482, RRG483, RRG484, RRG485, RRG486, RRG487, RRG488, RRG489, RRG490, RRG491, RRG492, RRG493, RRG494, RRG495, RRG496, RRG497, RRG498, RRG499, RRG500, RRG501, RRG502, RRG503, RRG504, RRG505, RRG506, RRG507, RRG508, RRG509, RRG510, RRG511, RRG512, RRG513, RRG514, RRG515, RRG516, RRG517, RRG518, RRG519, RRG520, RRG521, RRG522, RRG523, RRG524, RRG525, RRG526, RRG527, RRG528, RRG529, RRG530, RRG531, RRG532, RRG533, RRG534, RRG535, RRG536, RRG537, RRG538, RRG539, RRG540, RRG541, RRG542, RRG543, RRG544, RRG545, RRG546, RRG547, RRG548, RRG549, RRG550, RRG551, RRG552, RRG553, RRG554, RRG555, RRG556, RRG557, RRG558, RRG559, RRG560, RRG561, RRG562, RRG563, RRG564, RRG565, RRG566, RRG567, RRG568, RRG569, RRG570, RRG571, RRG572, RRG573, RRG574, RRG575, RRG576, RRG577, RRG578, RRG579, RRG580, RRG581, RRG582, RRG583, RRG584, RRG585, RRG586, RRG587, RRG588, RRG589, RRG590, RRG591, RRG592, RRG593, RRG594, RRG595, RRG596, RRG597, RRG598, RRG599, RRG600, RRG601, RRG602, RRG603, RRG604, RRG605, RRG606, RRG607, RRG608, RRG609, RRG610, RRG611, RRG612, RRG613, RRG614, RRG615, RRG616, RRG617, RRG618, RRG619, RRG620, RRG621, RRG622, RRG623, RRG624, RRG625, RRG626, RRG627, RRG628, RRG629, RRG630, RRG631, RRG632, RRG633, RRG634, RRG635, RRG636, RRG637, RRG638, RRG639, RRG640, RRG641, RRG642, RRG643, RRG644, RRG645, RRG646, RRG647, RRG648, RRG649, RRG650, RRG651, RRG652, RRG653, RRG654, RRG655, RRG656, RRG657, RRG658, RRG659, RRG660, RRG661, RRG662, RRG663, RRG664, RRG665, RRG666, RRG667, RRG668, RRG669, RRG670, RRG671, RRG672, RRG673, RRG674, RRG675, RRG676, RRG677, RRG678, RRG679, RRG680, RRG681, RRG682, RRG683, RRG684, RRG685, RRG686, RRG687, RRG688, RRG689, RRG690, RRG691, RRG692, RRG693, RRG694, RRG695, RRG696, RRG697, RRG698, RRG699, RRG700, RRG701, RRG702, RRG703, RRG704, RRG705, RRG706, RRG707, RRG708, RRG709, RRG710, RRG711, RRG712, RRG713, RRG714, RRG715, RRG716, RRG717, RRG718, RRG719, RRG720, RRG721, RRG722, RRG723, RRG724, RRG725, RRG726, RRG727, RRG728, RRG729, RRG730, RRG731, RRG732, RRG733, RRG734, RRG735, RRG736, RRG737, RRG738, RRG739, RRG740, RRG741, RRG742, RRG743, RRG744, RRG745, RRG746, RRG747, RRG748, RRG749, RRG750, RRG751, RRG752, RRG753, RRG754, RRG755, RRG756, RRG757, RRG758, RRG759, RRG760, RRG761, RRG762, RRG763, RRG764, RRG765, RRG766, RRG767, RRG768, RRG769, RRG770, RRG771, RRG772, RRG773, RRG774, RRG775, RRG776, RRG777, RRG778, RRG779, RRG780, RRG781, RRG782, RRG783, RRG784, RRG785, RRG786, RRG787, RRG788, RRG789, RRG790, RRG791, RRG792, RRG793, RRG794, RRG795, RRG796, RRG797, RRG798, RRG799, RRG800, RRG801, RRG802, RRG803, RRG804, RRG805, RRG806, RRG807, RRG808, RRG809, RRG810, RRG811, RRG812, RRG813, RRG814, RRG815, RRG816, RRG817, RRG818, RRG819, RRG820, RRG821, RRG822, RRG823, RRG824, RRG825, RRG826, RRG827, RRG828, RRG829, RRG830, RRG831, RRG832, RRG833, RRG834, RRG835, RRG836, RRG837, RRG838, RRG839, RRG840, RRG841, RRG842, RRG843, RRG844, RRG845, RRG846, RRG847, RRG848, RRG849, RRG850, RRG851, RRG852, RRG853, RRG854, RRG855, RRG856, RRG857, RRG858, RRG859, RRG860, RRG861, RRG862, RRG863, RRG864, RRG865, RRG866, RRG867, RRG868, RRG869, RRG870, RRG871, RRG872, RRG873, RRG874, RRG875, RRG876, RRG877, RRG878, RRG879, RRG880, RRG881, RRG882, RRG883, RRG884, RRG885, RRG886, RRG887, RRG888, RRG889, RRG890, RRG891, RRG892, RRG893, RRG894, RRG895, RRG896, RRG897, RRG898, RRG899, RRG900, RRG901, RRG902, RRG903, RRG904, RRG905, RRG906, RRG907, RRG908, RRG909, RRG910, RRG911, RRG912, RRG913, RRG914, RRG915, RRG916, RRG917, RRG918, RRG919, RRG920, RRG921, RRG922, RRG923, RRG924, RRG925, RRG926, RRG927, RRG928, RRG929, RRG930, RRG931, RRG932, RRG933, RRG934, RRG935, RRG936, RRG937, RRG938, RRG939, RRG940, RRG941, RRG942, RRG943, RRG944, RRG945, RRG946, RRG947, RRG948, RRG949, RRG950, RRG951, RRG952, RRG953, RRG954, RRG955, RRG956, RRG957, RRG958, RRG959, RRG960, RRG961, RRG962, RRG963, RRG964, RRG965, RRG966, RRG967, RRG968, RRG969, RRG970, RRG971, RRG972, RRG973, RRG974, RRG975, RRG976, RRG977, RRG978, RRG979, RRG980, RRG981, RRG982, RRG983, RRG984, RRG985, RRG986, RRG987, RRG988, RRG989, RRG990, RRG991, RRG992, RRG993, RRG994, RRG995, RRG996, RRG997, RRG998, RRG999, RRG1000	4 - 6 Wo	E
<b>Progressive externe Ophthalmoplegie mit mtDNA-Deletionen (PEOA, PEOB) *</b> 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
<b>Retinitis pigmentosa (RP), autosomal-dominant *</b> Gen-Panel: ID053.03, 29 Gene (64,9 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPG1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS, VWA8	3 - 5 Wo	E
<b>Retinitis pigmentosa (RP), autosomal-rezessiv *</b> Gen-Panel: ID050.04, 61 Gene (162,4 kb) ABCA4, AGLB5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513	4 - 6 Wo	E
<b>Retinitis pigmentosa (RP), umfassende Diagnostik *</b> Gen-Panel: ID288.02 Retinitis pigmentosa (RP), umfassende Diagnostik: 87 Gene (216,9 kb) ABCA4, AGLB5, 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, AGLB5, 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
<b>Senior-Loken-Syndrom (SLSN) *</b> Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E
<b>Septooptische Dysplasie *</b> Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROX1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
<b>Stickler-Syndrom (STL) *</b> Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Augenerkrankungen</b>		
<b>Usher-Syndrom (USH) *</b> 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
<b>Vitreoretinopathie *</b> Gen-Panel ID352.00 Vitreoretinopathie: 23 Gene (58,9 kb) ATOH7, BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CTNBN1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, PAK2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408 Exsudative Vitreoretinopathie (EVR): 15 Gene (34,9 kb) ATOH7, CAPN5, CTNBN1, 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
<b>Walker-Warburg-Syndrom (WWS, MDDGA) *</b> Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPBP, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
<b>Weill-Marchesani-Syndrom (WMS) *</b> Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
<b>Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD) *</b> Gen-Panel: ID101.03, 38 Gene (94,5 kb) ABCA4, ADAM9, AIPL1, ATF6, CACNA1F, CACNA2D4, CABP4, CDH3, CDHR1, CEP78, CEP250, CFAP418, CNGA3, CNGB3, CNNM4, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RP1, RPGR, RPGRIP1, SEMA4A, TLCD3B, TLL5, UNC119	3 - 5 Wo	E
<b>Bindegewebserkrankungen</b>		
<b>Bindegewebserkrankungen mit Aortenbeteiligung *</b> Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB3, TGFB3, 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, TGFB3, TGFB3, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFB2, TGFB3 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stickler-Syndrom (STL): 4 Gene (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 Wo	E
<b>Cutis laxa (ARCL, ADCL) *</b> Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
<b>Ehlers-Danlos-Syndrom (EDS) *</b> 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
<b>Hereditäre hämorrhagische Teleangiektasie (HHT) *</b> Gen-Panel: ID155.01, 7 Gene (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Bindegewebserkrankungen</b>		
<b>Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen *</b> 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, TGFB3, TGFB3, TGFB3, THSD4 Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3, TGFB3 Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB3, TGFB3, 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, TGFB3, TGFB3, TGFB3	4 - 6 Wo	E
<b>Marfan-Syndrom (MFS) *, #</b> Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFB3, TGFB3	2 - 4 Wo	E
<b>Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder *</b> Gen-Panel ID194.05 Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 48 Gene (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTS14, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan-Syndrom (MFS): 3 Gene (11,8 kb) FBN1, TGFB3, TGFB3 Loeys-Dietz-Syndrom (LDS) und Thorakales Aortenaneurysma (AAT): 17 Gene (42,6 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3, THSD4 Stickler-Syndrom (STL): 6 Gene (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	4 - 6 Wo	E
<b>Osteogenesis imperfecta (OI) *</b> Gen-Panel: ID066.02, 21 Gene (39,2 kb) ANOS, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEB38B, TENT5A, WNT1	3 - 5 Wo	E
<b>Stickler-Syndrom (STL) *</b> Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
<b>Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) *</b> 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, TGFB3, TGFB3, THSD4 Thorakales Aortenaneurysma, nicht-syndromale Form (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFB3, TGFB3, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3	3 - 5 Wo	E
<b>Endokrine Störungen</b>		
<b>Adipositas *</b> Gen-Panel: ID183.03 Adipositas: 57 Gene (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NROB2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 18 Gene (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NROB2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 40 Gene (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
<b>Adrenogenitales Syndrom (AGS, CAH) *</b> Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
<b>Diabetes insipidus *</b> Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Endokrine Störungen</b>		
<b>Endokrine Hypertonie *</b> Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoadosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDL): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E
<b>Glukokortikoid-Mangel (GCCD) *</b> Gen-Panel: ID222.00, 16 Gene (25,1 kb) AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NROB1, NR3C1, PCSK1, POMC, PROP1, STAR, TBX19, TXNRD2	3 - 5 Wo	E
<b>Hyperaldosteronismus (HALD) *</b> Gen-Panel: ID304.00, 6 Gene (20,6 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5	3 - 5 Wo	E
<b>Hyperinsulinämische Hypoglykämie (HHF) *</b> Gen-Panel: ID126.00, 8 Gene (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 Wo	E
<b>Hyperparathyreoidismus (HRPT) *</b> Gen-Panel: ID338.00, 10 Gene (19,2 kb) AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6	3 - 5 Wo	E
<b>Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung *</b> 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
<b>Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) *</b> 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
<b>Hypoparathyreoidismus *</b> 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
<b>Hypophysenadenom (PITA) *</b> Gen-Panel: ID387.00, 8 Gene (20,7 kb) AIP, CDH23, CDKN1B, GNAS, GPR101, MEN1, PRKAR1A, USP8	3 - 5 Wo	E
<b>Kongenitale Hypothyreose *</b> Gen-Panel: ID369.00 Kongenitale Hypothyreose: 37 Gene (69,0 kb) CDCAS8, DIO1, DUOX2, DUOX2, FOXE1, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, NKX2-1, NKX2-5, OTX2, PAX8, POU1F1, PRKAR1A, PROP1, RNPC3, ROBO1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TUBB1 Kongenitale Schilddrüsenunterfunktion ohne Kropf (CHNG): 8 Gene (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Schilddrüsen-Dyshormonogenese (TDH): 8 Gene (23,9 kb) DUOX2, DUOX2, IYD, SLC5A5, SLC26A4, SLC26A7, TG, TPO Kombinierter Hypophysenhormonmangel (CPHD): 8 Gene (11,9 kb) HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1	3 - 5 w	E
<b>Neonataler Diabetes mellitus *</b> Panel: ID162.01 Neonataler Diabetes mellitus: 29 Gene (53,6 kb) ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57 Permanenter neonataler Diabetes mellitus (PNDM): 10 Gene (15,9 kb) ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57 Syndromaler neonataler Diabetes mellitus: 21 Gene (40,5 kb) BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Endokrine Störungen</b>		
<b>Neuroendokrine Neoplasie *</b> Gen-Panel: ID386.00, 19 Gene (29,2 kb) AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL	3 - 5 Wo	E
<b>MODY-Diabetes *</b> 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
<b>Ovarialdysgenese (ODG) *</b> Gen-Panel: ID293.01 Ovarialdysgenese (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX-Gonadendysgenese (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX-Gonadendysgenese mit Taubheit (PRLTS): 6 Gene (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 Wo	E
<b>Phäochromozytom-Paragangliom-Syndrom (PPGL) *</b> Gen-Panel: ID042.03, 14 Gene (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E
<b>Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) *</b> Gen-Panel: ID250.01 Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA): 8 Gene (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 Wo	E
<b>Schilddrüsenkarzinom *</b> Gen-Panel: ID220.02, 26 Gene (60,5 kb) ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53	3 - 5 Wo	E
<b>Septooptische Dysplasie *</b> Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
<b>Störung der Geschlechtsentwicklung (DSD) *</b> Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NROB1, NR2F2, NR5A1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NROB1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSP01, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NROB1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
<b>Vorzeitige Ovarialinsuffizienz (POF) *</b> 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
<b>Wachstumshormonmangel (IGHD, CPHD) *</b> Gen-Panel: ID211.02 Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNPC3, OTX2, POU1F1, PROP1, SEMA3A, SOX3 Isolierter Wachstumshormonmangel (IGHD): 5 Gene (6,4 kb) BTK, GH1, GHRHR, GHSR, SOX3 Kombinierter Hypophysenhormonmangel (CPHD): 10 Gene (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Entwicklungs- und Wachstumsstörungen</b>		
<b>Adipositas *</b> Gen-Panel: ID183.03 Adipositas: 57 Gene (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NROB2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Adipositas, nicht syndromal: 18 Gene (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NROB2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Adipositas, syndromal: 40 Gene (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 Wo	E
<b>Autismus-Spektrum-Störungen *</b> Gen-Panel: ID076.04, 168 Gene (664,6 kb) AHDC1, ADNP, ADSL, AFF2, AGO1, ALDH5A1, ANK2, ANKRD11, ARHGEF9, ARID1B, ARX, ASH1L, ASTN2, ASXL3, ATP1A1, AUTS2, BAZ2B, BCKDK, BCL11A, BRSK2, CACNA1C, CAPRIN1, CASK, CASZ1, CC2D1A, CDKL5, CELF4, CHD2, CHD7, CHD8, CIC, CNOT3, CNTN4, CNTNAP2, CREBBP, CSDE1, CSNK2A1, CTNNA1, CUL3, DDX3X, DEAF1, DHCR7, DIP2A, DLGAP2, DNMT3A, DPP6, DSCAM, DYNC1H1, DYRK1A, EBF3, EHMT1, EIF4E, EP300, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, GIGYF1, GLRA2, GRIA2, GRIN2A, GRIN2B, HRC2, HNRNPU, IL1RAPL1, IQSEC2, IRF2BPL, KATNAL2, KCNQ3, KDM5A, KDM5C, KDM6B, KMT2A, KMT2C, KMT2E, KMT5B, MAGEL2, MAOA, MBD5, MBOAT7, MECP2, MED13, MED13L, MEF2C, MEIS2, MYT1L, NAA15, NEXMIF, NF1, NHS, NIPBL, NLGN1, NLGN3, NLGN4X, NOVA2, NR1I3, NR4A2, NRXN1, NRXN2, NRXN3, NSD1, OPHN1, PAH, PAX5, PCDH19, PDZD8, PHF12, PHF2, PHF21A, PHF3, POGZ, PPP2R5D, PSMD12, PTCHD1, PTEN, RAB39B, RAI1, RELN, RERE, RFX3, RIMS1, RORB, RPL10, SATB2, SCN1A, SCN2A, SEMA5A, SETD2, SETD5, SGSH, SHANK2, SHANK3, SLC6A1, SLC6A8, SLC9A6, SLC9A9, SMARCB1, SMARCC2, SON, SOX5, SPAST, STXBP1, SYN1, SYNGAP1, TANC2, TBL1XR1, TBR1, TCF20, TCF4, TLK2, TMLHE, TRIP12, TRPC6, TRRAP, TSC1, TSC2, UBE3A, UPF3B, VAMP2, VPS13B, WAC, WDFY3, ZMYM3, ZMYND8, ZNF292, ZNF462, ZSWIM6	4 - 6 Wo	E
<b>Coffin-Siris-Syndrom (CSS) *</b> Gen-Panel: ID118.01, 14 Gene (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 Wo	E
<b>Cornelia-de-Lange-Syndrom (CDLS) *</b> Gen-Panel: ID033.02, 8 Gene (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 Wo	E
<b>FG-Syndrom (FGS) *</b> Gen-Panel: ID215.00, 3 Gene (17,2 kb) CASK, FLNA, MED12	3 - 5 Wo	E
<b>Großwuchs, umfassende Diagnostik *</b> Gen-Panel: ID299.01, 64 Gene (211,5 kb) ABCC9, AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, FBN1, FIBP, GFAP, GLI3, GPC3, GPC4, GRIA3, H1-4, HEPACAM, HERC1, HUWE1, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIGA, PIK3CA, PIK3R2, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SHANK, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMEM94, TRIP12, UPF3B, ZBTB20, ZDHHC9	4 - 6 Wo	E
<b>Großwuchssyndrome (SOTOS, BWS) *</b> Gen-Panel: ID073.05 Großwuchssyndrome (SOTOS, BWS): 15 Gene (41,2 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, PDGFRB, PTEN, SETD2, SUZ12, RNF125 Sotos-ähnliche Großwuchssyndrome: 6 Gene (23,1 kb) EED, EZH2, NFIX, NSD1, SETD2, SUZ12 Beckwith-Wiedemann-ähnliche Großwuchssyndrome: 5 Gene (10,1 kb) CDKN1C, DIS3L2, GPC3, GPC4, OFD1	3 - 5 Wo	E
<b>Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *</b> Gen-Panel: ID292.00, 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
<b>Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD, IDD) *</b> Gen-Panel: ID036.06 Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD, IDD): 105 Gene (354,6 kb) ACTL6B, ADNP, AHDC1, AP2M1, ARID1A, ARID1B*, ARID2, ASH1L, ATP2B1, AUTS2, BCL11A, BCL11B, BICRA, BRPF1, CACNG2, CAMK2A, CAMK2B, CAMK2G, CCNK, CDH15, CDK8, CERT1, CHAMP1, CHD3, CHD8, CIC, CLTC, CNOT2, CNOT3, CTCF, CTNNA1, DDX6, DEAF1, DHX9, DLG4, DPF2, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, FBXO11, FOXP1, GATAD2B, GNB1, GRIA1, GRIN1, GRIN2B, HIVEP2, HNRNPC, KAT6A, KCNQ5, KDM3B, KDM4B, KIF1A, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MEF2C, MTSS2, MYT1L, NAA15, NR4A2, NUS1, PACS1, PAK1, PHF21A, POGZ, PPP2R1A, PPP2R5D, PURA*, RAC1, RFX7, RORA, SET, SETBP1, SETD1B, SETD2, SETD5, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4, SOX6, SRRM2, STAG1, SYNGAP1, TAF4, TANC2, TBL1XR1, TBR1, TCF1, TLK2, TNPO2, TRIO, TRIP12, USP7, ZBTB18, ZMYND11, ZNF292 Intellektuelle Entwicklungsstörung, autosomal-dominant, nicht-syndromal (MRD): 62 Gene (224,4 kb) AP2M1, ARID1A, ARID1B, ASH1L, ATP2B1, AUTS2, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CIC, CLTC, CTCF, DEAF1, DHX9, DLG4, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN2B, HIVEP2, HNRNPC, KAT6A, KCNQ5, KDM4B, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, RAC1, RFX7, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SOX11, SRRM2, STAG1, SYNGAP1, TAF4, TBL1XR1, TLK2, TRIO, ZBTB18, ZMYND11, ZNF292 Intellektuelle Entwicklungsstörung, autosomal-dominant, syndromal (IDD): 55 Gene (168,0 kb) ACTL6B, ADNP, AHDC1, ARID1A, ARID1B, ARID2, BCL11A, BCL11B, BICRA, BRPF1, CCNK, CDK8, CERT1, CHAMP1, CHD3, CHD8, CNOT2, CNOT3, CTNNA1, DDX6, DEAF1, DPF2, FBXO11, FOXP1, GATAD2B, GRIN1, KAT6A, KDM3B, KIF1A, MEF2C, MTSS2, NR4A2, PACS1, PAK1, PHF21A, POGZ, PPP2R1A, PPP2R5D, PURA, RORA, SETD1B, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4, SOX6, TANC2, TBR1, TCF1, TNPO2, TRIP12, USP7	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Entwicklungs- und Wachstumsstörungen</b>		
<b>Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT, IDD) *</b> Gen-Panel: ID037.03 Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT, IDD): 77 Gene (201,7 kb) ABCA2, ACTL6B, ADAT3, ALG14, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CPE, CRADD, CRBN, DEAF1, EDC3, EIF3F, ELP2, FBXL3, FBXO31, FERRY3, FMN2, GNB5, GRIA1, GRIK2, HERC2, HNMT, IMPA1, IQSEC1, KDM5B, KPTN, LIG3, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NEMF, NSUN2, NSUN6, NUDT2, OTUD6B, PDE2A, PDZD8, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, PUS7, RSRC1, RUSC2, SCAPER, SLC45A1, SLC6A17, ST3GAL3, TAF13, TAF2, TECR, TMEM94, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, WIPI2, ZBTB11, ZC3H14 Intellektuelle Entwicklungsstörung, autosomal-rezessiv, nicht-syndromal (MRT): 58 Gene (157,9 kb) ADAT3, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31, FERRY3, FMN2, GRIA1, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NSUN2, NSUN6, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, ZBTB11, ZC3H14 Intellektuelle Entwicklungsstörung, autosomal-rezessiv, syndromal (IDD): 20 Gene (48,5 kb) ABCA2, ALG14, CPE, DEAF1, FBXL3, GNB5, IQSEC1, LIG3, NEMF, NUDT2, OTUD6B, PDE2A, PDZD8, PUS7, SCAPER, SLC45A1, TAF2, TMEM94, WIPI2, ZBTB11	4 - 6 Wo	E
<b>Intellektuelle Entwicklungsstörung, X-chromosomal (XLID, MRX, MRXS) *</b> 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, SLC16A2, SLC9A6, SMS, TAF1, UBE2A, UPF3B, USP9X, ZC4H2, ZDHHC9	4 - 6 Wo	E
<b>Intellektuelle Entwicklungsstörung und Makrozephalie *</b> Gen-Panel: ID131.03 Intellektuelle Entwicklungsstörung und Makrozephalie: 48 Gene (147,3 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMCO1, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9 Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Makrozephalie: 21 Gene (70,8 kb) CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20 Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Makrozephalie: 10 Gene (20,2 kb) ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMCO1, ZBTB7A Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Makrozephalie: 18 Gene (57,9 kb) BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9	4 - 6 Wo	E
<b>Intellektuelle Entwicklungsstörung und Mikrozephalie *</b> Gen-Panel: ID129.02 Intellektuelle Entwicklungsstörung und Mikrozephalie: 80 Gene (229,8 kb) ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNBN1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, 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, CTNBN1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2 Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Mikrozephalie: 21 Gene (41,0 kb) ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73 Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Mikrozephalie: 27 Gene (88,3 kb) ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2	4 - 6 Wo	E
<b>Neurologische Entwicklungsstörungen (NED), umfassende Diagnostik *</b> Gen-Panel: ID358.00, 169 Gene (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6VOA1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNBN1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDTF1, 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, RALGAP1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMP1, TIAM1, TMEM147, TMEM222, TMX2, TNF, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Entwicklungs- und Wachstumsstörungen</b>		
<b>Kabuki-Syndrom (KABUK) *, #</b> Gen-Panel: ID127.00, 2 Gene (20,8 kb) KDM6A, KMT2D	3 - 5 Wo	E
<b>Kleinwuchs, umfassende Diagnostik *</b> 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, ERCC6, ERCC8, EXOC6B, EXOSC2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGD1, FGFR3, FLNB, FN1, GALNS, GDF5, GH1, GHR, GHRHR, GHSR, GLB1, GLI2, GMNN, GNPAT, GPX4, GRHL2, GSC, GUSB, GZF1, HDAC8, HESX1, HMGA2, HRAS, HYAL1, IDUA, IGF1, IGF1R, IGF2, IGFALS, INSR, IRS1, IRS4, KIF22, KMT2A, KRAS, LARP7, LFNG, LHX3, LHX4, LTBP3, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MCM5, MRAS, NBAS, NBN, NEPRO, , NIPBL, NKX2-5, NKX3-2, NOTCH2, NPR2, NRAS, NSMCE2, OBSL1, ORC1, ORC4, ORC6, OTX2, PALB2, PAM16, PAPP2, PAX8, PCNT, PEX5, PEX7, PHEX, PIK3R1, PISD, PLAG1, PLCB3, POC1A, POLR3GL, POP1, POU1F1, PPP1CB, PPP1R15B, PPP3CA, PRKG2, PRMT7, PROP1, PTH1R, PTPN11, PUS7, RAD21, RAD51, RAD51C, RAF1, RBBP8, RFW3, RIT1, RMRP, 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, TRAI, TRAPPC2, TRHR, TRIM37, TRIP11, TRMT10A, , TRPV4, TSHB, TSHR, UBE2T, XRCC2, XRCC4	4 - 6 Wo	E
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, TRAI 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, PAPS2, PISD, PLCB3, RMRP, RNU4ATAC, POP1, PRKG2, RPL13, RSPRY1, SIK3, SLC26A2, TONSL, TRAPPC2, TRIP11, TRPV4		
<b>Noonan-Syndrom (NS) *</b> 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
<b>Pitt-Hopkins-Syndrom (PTHS) *</b> Gen-Panel: ID106.00, 3 Gene (10,6 kb) CNTNAP2, NRXN1, TCF4	3 - 5 Wo	E
<b>Progerie und progeroide Syndrome *</b> Gen-Panel: ID147.01, 25 Gene (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 Wo	E
<b>RASopathien *</b> 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 Kardiofaziokutanenes 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
<b>Rett-Syndrom (RTT) und ähnliche Krankheitsbilder *</b> Gen-Panel: ID125.01, 16 Gene (45,3 kb) CDKL5, FOXP1, GABBR2, GABRA2, GABRB2, GRIN2B, HTT, IQSEC2, MECP2, MEF2C, NTNG2, SHANK3, STXBP1, TCF4, UBE3A, WDR45	3 - 5 Wo	E
<b>Rubinstein-Taybi-Syndrom (RSTS) *</b> Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
<b>Seckel-Syndrom (SCKL) *</b> Gen-Panel: ID113.00, 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAI	3 - 5 Wo	E
<b>Sprachentwicklungsstörung (DLD, CAS) *</b> 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, ZFH4, 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, ZFH4, ZNF142 Spezifische Sprachbeeinträchtigung (SLI): 5 Gene (9,5 kb) TP2C2, BUD13, NFXL1, TM4SF20, ZNF277	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Entwicklungs- und Wachstumsstörungen</b>		
<b>Wachstumshormonmangel (IGHD, CPHD) *</b> Gen-Panel: ID211.02 Wachstumshormonmangel (IGHD, CPHD): 15 Gene (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, RNP3, 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, RNP3, SEMA3A	3 - 5 Wo	E
<b>Wachstumsstörung und Makrozephalie *</b> 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
<b>3M-Syndrom *</b> Gen-Panel: ID214.00, 3 Gene (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 Wo	E
<b>Epilepsien und Migräne</b>		
<b>Absence-Epilepsie (EJA, ECA) *</b> Gen-Panel: ID057.02, 10 Gene (21,8 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, KCNMA1, RORB, SLC2A1, SLC12A5	3 - 5 Wo	E
<b>Benigne neonatale und infantile Krampfanfälle (BFNS, BFIS) *</b> Gen-Panel: ID134.01, 6 Gene (19,8 kb) CHRNA2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A	3 - 5 Wo	E
<b>Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE) *</b> Gen-Panel ID080.03 Entwicklungsbedingte und epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGFE9, ARV1, ARX, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNMT1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOB2, 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, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CDK19, CELF2, CHD2, CUX2, CYFIP2, DNMT1, EEF1A2, FBXO28, FGF12, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GNAO1, GRIN2B, GRIN2D, HCN1, HNRNPU, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, NEUROD2, NSF, NTRK2, PACS2, PHACTR1, PPP3CA, RHOB2, 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, ARHGFE9, ARX, CDKL5, FGF13, PCDH19, PIGA, SLC35A2, SMC1A	4 - 6 Wo	E
<b>Epileptische Enzephalopathien, umfassende Diagnostik *</b> Gen-Panel ID047.04 Epileptische Enzephalopathien, umfassende Diagnostik: 163 Gene (371,9 kb) AARS1, ABAT, ACTL6B, ADAM22, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP3B2, ARHGFE9, ARV1, ARX, ASNS, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, BRAT1, BSCL2, BTD, CACNA1A, CACNA1E, CAD, CCDC88A, CDK19, CDKL5, CELF2, CHD2, CLCN4, CNPY3, CPLX1, CNTNAP2, CUX2, CYFIP2, D2HGDH, DALRD3, DENND5A, DNMT1, DNMT1L, DMXL2, DOCK7, EEF1A2, ETHE1, FGF12, FGF13, FOLR1, FOXG1, FBXO28, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, 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, RHOB2, 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, 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, ARHGFE9, ARV1, ARX, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNMT1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOB2, 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

Erkrankung/Diagnostik	Dauer	Material
<b>Epilepsien und Migräne</b>		
<b>Epilepsien, umfassende Diagnostik *</b> Gen-Panel ID061.05 Epilepsien, umfassende Diagnostik: 163 Gene (388,9 kb) AARS1, ACTL6B, ADAM22, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ASAH1, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, BRAT1, CACNA1A, CACNA1E, CACNB4, CAD, CASR, CDK19, CDKL5, CELF2, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CILK1, CLCN2, CLN8, CNPY3, CNTN2, CNTNAP2, CPA6, CPLX1, CSTB, CUX2, CYFIP2, DALRD3, DENND5A, DEPDC5, DMXL2, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GLS, GNAO1, GOT2, GOSR2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GUF1, HCN1, HCN2, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC1, KCNC2, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LGI1, LMNB2, MDH1, MDH2, MECP2, MEF2C, MTOR, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PLPBP, PPP3CA, PRDM8, PRICKLE1, PRRT2, RELN, RHOB2, RNF13, ROGD1, RORA, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC38A3, SLC6A1, SLC7A6OS, SMC1A, SNIP1, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Fokale Epilepsien: 17 Gene (50,9 kb) CHRNA2, CHRNA4, CHRN2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24 Generalisierte Epilepsien: 39 Gene (72,6 kb) ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A6OS, SLC12A5, STX1B, TBC1D24 Epileptische Enzephalopathie (DEE, EIEE): 105 Gene (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP*, PPP3CA, RHOB2, 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
<b>Epilepsie mit schwerer Entwicklungsstörung *</b> 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
<b>Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFS) *</b> Gen-Panel: ID059.03 Fieberkrämpfe mit oder ohne Epilepsie (FEB, GEFS): 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 (GEFS): 7 Gene (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 Wo	E
<b>Fokale Epilepsien *</b> Gen-Panel: ID208.01 Fokale Epilepsien: 17 Gene (50,9 kb) CHRNA2, CHRNA4, CHRN2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24 Fokale Epilepsie mit variablen Herden (FFEVF): 4 Gene (13,7 kb) DEPDC5, NPRL2, NPRL3, SCN3A Nächtliche Frontallappenepilepsie (ENFL): 5 Gene (13,5 kb) CHRNA2, CHRNA4, CHRN2, DEPDC5, KCNT1 Temporallappenepilepsie (ETL): 4 Gene (13,4 kb) CPA6, GAL, LGI1, RELN	3 - 5 Wo	E
<b>Generalisierte Epilepsien *</b> Gen-Panel: ID040.05 Generalisierte Epilepsien: 39 Gene (72,6 kb) ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A6OS, SLC12A5, STX1B, TBC1D24 Myoklonusepilepsie (EJM, EPM): 24 Gene (45,3 kb) ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6B, SLC7A6OS, TBC1D24 Absence-Epilepsie (EJA, ECA): 9 Gene (18,3 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, RORB, SLC2A1, SLC12A5 Generalisierte Epilepsie mit Fieberkrämpfen (GEFS): 7 Gene (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 Wo	E
<b>Generalisierte Epilepsie mit Fieberkrämpfen plus (GEFS, GEFS+) *</b> Gen-Panel: ID235.03, 8 Gene (17,2 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, SLC32A1, STX1B	3 - 5 Wo	E
<b>Migräne (MGR) *</b> 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

Erkrankung/Diagnostik	Dauer	Material
<b>Epilepsien und Migräne</b>		
<b>Hemiplegische Migräne (FHM) *</b> Gen-Panel: ID064.02, 7 Gene (23,1 kb) ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1	3 - 5 Wo	E
<b>Metabolische Epilepsien *</b> Gen-Panel: ID303.01 Metabolische Epilepsien: 84 Gene (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycin-Enzephalopathie (GCE): 5 Gene (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdän-Cofaktor-Defizienz (MOCOD): 3 Gene (3,7 kb) GPHN, MOCS1, MOCS2 Zerebrales Kreatinmangelsyndrom (CCDS): 3 Gene (3,9 kb) GAMT, GATM, SLC6A8 Ahornsirupkrankheit (MSUD): 5 Gene (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxy-Glutarazidurie: 3 Gene (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-Gangliosidose: 3 Gene (3,8 kb) HEXA, HEXB, GM2A Neuronale Ceroid-Lipofuszinose (CLN): 12 Gene (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 Wo	E
<b>Myoklonusepilepsie (EPM, EJM) *</b> 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
<b>Stoffwechselstörung mit Epilepsie im Neugeborenenalter *</b> 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
<b>Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter *</b> 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
<b>Stoffwechselstörung mit Epilepsie im Schul- und Jugendalter *</b> 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
<b>Fertilitätsstörungen</b>		
<b>Adrenogenitales Syndrom (AGS, CAH) *</b> Gen-Panel: ID111.02, 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 Wo	E
<b>Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) *</b> 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
<b>Männliche Infertilität (SPGF, CBAVD) *</b> Gen-Panel ID192.05 Männliche Infertilität (SPGF, CBAVD): 108 Gene (368,8 kb) ACR, ACTL7A, ACTL9, ADGRG2, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP251, CFAP43, CFAP44, CFAP47, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFTR, CT55, CYLC1, DMRT1, DNAH1, DNAH10, DNAH17, DNAH2, DNAH8, DNALI1, DNHD1, DOPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRR23, M1AP, MEI1, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Störung der Spermatogenese (SPGF): 102 Gene (349,2 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP251, CFAP43, CFAP44, CFAP47, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CT55, CYLC1, DNAH1, DNAH10, DNAH17, DNAH2, DNAH8, DNALI1, DNHD1, DOPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, IFT74, IQCN, KASH5, KCNU1, KLHL10, LRR23, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Kongenitale bilaterale Aplasie des Vas deferens (CBAVD): 2 Gene (7,5 kb) ADGRG2, CFTR	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Fertilitätsstörungen</b>		
<b>Ovarialdysgenese (ODG) *</b> Gen-Panel: ID293.01 Ovarialdysgenese (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX-Gonadendysgenese (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX-Gonadendysgenese mit Taubheit (PRLTS): 6 Gene (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 Wo	E
<b>Perrault-Syndrom (PRLTS) *</b> Gen-Panel: ID388.00, 9 Gene (14,1 kb) CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, MRPL49, PRORP, TWNK	3 - 5 Wo	E
<b>Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *</b> Gen-Panel: ID085.03, 50 Gene (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 Wo	E
<b>Störung der Geschlechtsentwicklung (DSD) *</b> Gen-Panel: ID117.03 Störung der Geschlechtsentwicklung (DSD): 49 Gene (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NROB1, NR2F2, NR5A1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Störung der Geschlechtsentwicklung (SRXY), nicht-syndromal: 21 Gene (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NROB1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Störung der Geschlechtsentwicklung (SRXX), nicht-syndromal: 6 Gene (6,6 kb) NR2F2, NR5A1, RSP01, SOX9, SRY, WNT4 Störung der Geschlechtsentwicklung (DSD), syndromal: 37 Gene (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NROB1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenitales Syndrom (AGS, CAH): 7 Gene (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 Wo	E
<b>Störung der Oozyten-, Zygoten- und Embryonen-Reifung (OZEMA) *</b> Gen-Panel: ID239.02, 22 Gene (36,7 kb) ASTL, BTG4, CDC20, CHEK1, FBXO43, KPNA7, MOS, NLRP2, NLRP5, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3	3 - 5 Wo	E
<b>Vorzeitige Ovarialinsuffizienz (POF) *</b> 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
<b>Fettstoffwechselstörungen</b>		
<b>Fettstoffwechselstörungen, umfassende Diagnostik *</b> Gen-Panel: ID044.03, 62 Gene (114,6 kb) ABCA1, ABCG5, ABCG8, ACADM, ACADS, ACADVL, ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA1, APOA2, APOA4, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CETP, CIDEA, CREB3L3, CYP27A1, CYP7A1, DHCR7, DHCR24, GCKR, GK, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIMA1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPA, LPL, MTP, NPC1, NPC1L1, NPC2, PCSK9, PCYT1A, PLAAT3, PLIN1, PNPLA5, POLD1, PPARG, SAR1B, SCARB1, SMPD1, SORT1, ZMPSTE24	4 - 6 Wo	E
<b>Fettstoffwechselstörung durch LDL-Mangel *</b> Gen-Panel: ID094.01, 10 Gene (29,6 kb) ANGPTL3, ANGPTL4, ANGPTL8, APOB, APOE, LIMA1, MTP, NPC1L1, PCSK9, SAR1B	3 - 5 Wo	E
<b>HDL-assoziierte Fettstoffwechselstörung *</b> 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
<b>Hypertriglyceridämie *</b> Gen-Panel: ID095.03 Hypertriglyceridämie: 32 Gene (51,1 kb) ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24 Hyperchylomikronämie: 6 Gene (6,1 kb) APOC2, APOE, APOA5, GPIHBP1, LMF1, LPL Lipodystrophie: 15 Gene (22,0 kb) ADRA2A, AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEA, LIPE, LMNA, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Fettstoffwechselstörungen</b>		
<b>Hypercholesterinämie *</b> 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
<b>Kombinierte Hyperlipidämie *</b> Gen-Panel: ID025.06 Kombinierte Hyperlipidämie: 40 Gene (72,8 kb): ABCG5, ABCG8, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, CREB3L3, CYP27A1, CYP7A1, GCKR, GK, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, NPC1L1, PCSK9, PLIN1, PNPLA5, POLD1, PPARG, SORT1, ZMPSTE24 Hypercholesterinämie: 13 Gene (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1 Hypertriglyceridämie: 29 Gene (51,1 kb) AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PLIN1, POLD1, PPARG, ZMPSTE24	3 - 5 Wo	E
<b>Lipodystrophie (CGL, FPLD) *</b> Gen-Panel: ID055.04 Lipodystrophie (CGL, FPLD): 15 Gene (22,0 kb) ADRA2A, AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEA, LIPE, LMNA, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24 Kongenitale generalisierte Lipodystrophie (CGL): 5 Gene (4,9 kb) AGPAT2, BSCL2, CAV1, CAVIN1, PCYT1A Familiäre partielle Lipodystrophie (FPLD): 8 Gene (11,5 kb) ADRA2A, CAV1, CIDEA, LIPE, LMNA, PLAAT3, PLIN1, PPARG	3 - 5 Wo	E
<b>Lipodystrophien, umfassende Diagnostik *</b> Gen-Panel: ID343.00, 45 Gene (80,0 kb) ADRA2A, AGPAT2, AKT2, ALDH18A1, BANF1, BSCL2, CAV1, CAVIN1, CIDEA, EPHX1, ERCC6, ERCC8, FBN1, HRAS, KCNJ6, LEP, LEPR, LIPE, LMNA, MCM3, MCM7, MFN2, MTX2, NSMCE2, OPA3, OTULIN, PCYT1A, PDGFRB, PIK3R1, PLIN1, POLD1, POLR3A, POMP, PPARG, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PTPN11, SLC25A24, SLC29A3, SPRN, WRN, ZMPSTE24	3 - 5 Wo	E
<b>Statin-assoziierte Myopathie *</b> Gen-Panel: ID097.00, 11 Gene (37,2 kb) ACADM, ACADS, ACADVL, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1, SLC01B1	3 - 5 Wo	E
<b>Gastroenterologische Erkrankungen</b>		
<b>Alagille-Syndrom (ALGS) *, #</b> Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
<b>Cholestase, umfassende Diagnostik *</b> 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
<b>Gastrointestinaler Stromatumor (GIST) *</b> Gen-Panel: ID226.00, 8 Gene (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 Wo	E
<b>Lynch-Syndrom (LYNCH, HNPCC) *</b> Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	2 - 4 Wo	E
<b>Intrahepatische Cholestase (PFIC, CBAS) *</b> Gen-Panel: ID159.04 Intrahepatische Cholestase (PFIC, CBAS): 25 Gene (64,9 kb) ABCB4, ABCB11, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, AKR1D1, AMACR, ATP8B1, BAAT, CYP7B1, HSD3B7, JAG1, KIF12, MYO5B, NOTCH2, NR1H4, SEMA7A, SLC25A13, SLC51A, TJP2, USP53, VPS33B, ZFYVE19 Progressive intrahepatische Cholestase (PFIC): 13 Gene (35,2 kb) ABCB4, ABCB11, ABCG8, ATP8B1, KIF12, MYO5B, NR1H4, SEMA7A, SLC51A, TJP2, USP53, VPS33B, ZFYVE19 Gallensäuresynthesedefekt (CBAS): 7 Gene (10,0 kb) ABCD3, ACOX2, AKR1D1, AMACR, BAAT, CYP7B1, HSD3B7 Alagille-Syndrom (ALGS): 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
<b>Kolorektales Karzinom und Polyposis *</b> Gen-Panel ID006.09, 23 Gene (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53	3 - 5 Wo	E
<b>Magenkarzinom *</b> Gen-Panel: ID090.04, 25 Gene (73,6 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	3 - 5 Wo	E
<b>Morbus Hirschsprung (HSCR) *</b> 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

Erkrankung/Diagnostik	Dauer	Material
<b>Gastroenterologische Erkrankungen</b>		
<b>Pankreaskarzinom *</b> Gen-Panel: ID089.03, 17 Gene (55,2 kb) APC, ATM, BRCA1, BRCA2, CDKN2A, CTSC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL	3 - 5 Wo	E
<b>Pankreatitis (PCTT) *</b> Gen-Panel: ID141.02, 18 Gene (29,6 kb) APOA5, APOC2, CASR, CEL, CFTR, CPA1, CLDN2, CELA3B, CTSC, CTSB, GPIHBP1, LMF1, LPL, PNLIP, PRSS1, SPINK1, TRPV6, UBR1	3 - 5 Wo	E
<b>Polyposis-Syndrom (PS, FAP) *</b> Gen-Panel: ID005.06, 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
<b>Polyzystische Lebererkrankung (PCLD) *</b> Gen-Panel: ID305.02 Polyzystische Lebererkrankung (PCLD): 16 Gene (55,7 kb) ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63 Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 7 Gene (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 9 Gene (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1	3 - 5 Wo	E
<b>Viszerale Myopathien und Neuropathien, umfassende Diagnostik *</b> 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
<b>Gehirnfehlbildungen</b>		
<b>Aicardi-Goutières-Syndrom (AGS) *</b> Gen-Panel: ID058.01, 9 Gene (13,0 kb) ADAR, IFIH1, LSM11, RNASEH2A, RNASEH2B, RNASEH2C, RNU7-1, SAMHD1, TREX1	3 - 5 Wo	E
<b>Holoprosenzephalie (HPE) *</b> 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
<b>Joubert-Syndrom (JBTS) *</b> Gen-Panel: ID028.03, 40 Gene (104,1 kb) AH11, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 Wo	E
<b>Komplexe kortikale Dysplasie mit weiteren Hirnfehlbildungen (CDCBM) *</b> Gen-Panel: ID271.02, 15 Gene (52,0 kb) ADGRG1, APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2	3 - 5 Wo	E
<b>Lissenzephalie (LIS) *</b> 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
<b>Meckel-Syndrom (MKS) *</b> 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
<b>Neuronale Migrationsstörungen, umfassende Diagnostik *</b> Gen-Panel: ID180.01 Neuronale Migrationsstörungen, umfassende Diagnostik: 82 Gene (264,4 kb) ACTB, ACTG1, ADGRG1, AKT3, APC2, ARF1, ARFGF2, 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, ARFGF2, 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 Schlitzenzephalie: 7 Gene (19,7 kb) COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Gehirnfehlbildungen</b>		
<b>Periventrikuläre noduläre Heterotopie (PVNH) *</b> Gen-Panel: ID306.00, 6 Gene (26,1 kb) ARF1, ARFGEF2, ERMARD, FLNA, MAP1A, NEDD4L	3 - 5 Wo	E
<b>Polymikrogyrie *</b> 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
<b>Pontocerebelläre Hypoplasie (PCH) *</b> 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
<b>Schizenzephalie *</b> Gen-Panel: ID173.00, 7 Gene (19,7 kb) COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62	3 - 5 Wo	E
<b>Septooptische Dysplasie *</b> Gen-Panel: ID378.00, 8 Gene (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROX1, SOX2, SOX3, TAX1BP3	3 - 5 Wo	E
<b>Walker-Warburg-Syndrom (WWS, MDDGA) *</b> Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKR, FKTN, GMPBP, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
<b>Zerebelläre Dysgenese, X-chromosomal *</b> Gen-Panel: ID219.00, 11 Gene (24,5 kb) ABC7, CASK, DKC1, FMR1, L1CAM, MECP2, MID1, OFD1, OPHN1, SLC9A6, ZIC3	3 - 5 Wo	E
<b>Hämatologische Erkrankungen</b>		
<b>Atypisches hämolytisch-urämisches Syndrom (AHUS) *</b> 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
<b>Erythrozytose (ECYT) *</b> Gen-Panel: ID138.02, 11 Gene (13,9 kb) BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL	3 - 5 Wo	E
<b>Fanconi-Anämie (FANC) *</b> Gen-Panel: ID043.02, 21 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFW3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E
<b>Hämophilie *</b> Gen-Panel: ID154.01, 5 Gene (18,9 kb) F8, F9, LMAN1, MCFD2, VWF	3 - 5 Wo	E
<b>Hermansky-Pudlak-Syndrom (HPS) *</b> Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
<b>Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) *</b> 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, EPICAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GF11, 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, RFW3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCHC8 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, ZCHC8 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, GF11, 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, RFW3, SLX4, UBE2T, XRCC2 Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb) MLH1, MSH2, MSH6, PMS2	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Hämatologische Erkrankungen</b>		
<b>Knochenmarkinsuffizienz (BMF) *</b> Gen-Panel: ID357.00 Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb): ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTE11, SRP72, TERC, TERT, TP53, ZCCHC8 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTE11, TERC, TERT, ZCCHC8	3 - 5 Wo	E
<b>Neutropenie *</b> Gen-Panel: ID189.02 Neutropenie: 30 Gene (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndrome mit Neutropenie: 21 Gene (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, USB1, VPS13B, WDR1	3 - 5 Wo	E
<b>Porphyrrie *</b> Gen-Panel: ID153.01, 10 Gene (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 Wo	E
<b>Sideroblastische Anämie (SIDBA) *</b> Gen-Panel: ID355.00, 10 Gene (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2	3 - 5 Wo	E
<b>Sphärozytose (SPH) und Elliptozytose (EL) *</b> Gen-Panel: ID203.01 Sphärozytose (SPH) und Elliptozytose (EL): 6 Gene (26,8 kb) ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB Sphärozytose (SPH): 5 Gene (24,2 kb) ANK1, EPB42, SLC4A1, SPTA1, SPTB Elliptozytose (EL): 4 Gene (18,4 kb) EPB41, SLC4A1, SPTA1, SPTB	3 - 5 Wo	E
<b>Thrombophilie (THPH) *</b> Gen-Panel: ID150.01, 12 Gene (25,2 kb) F2, F5, F9, F13B, HABP2, HRG, MTHFR, PROC, PROS1, SERPINC1, SERPIND1, THBD	3 - 5 Wo	E
<b>Thrombozytendefekte, umfassende Diagnostik *</b> Gen-Panel: ID274.01 Thrombozytendefekte, umfassende Diagnostik: 64 Gene (142,3 kb) ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, EPHB2, ETV6, FERMT3, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB3, JAK2, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TUBB1, WAS, WDR1, WIPF1 Thrombozytopenie, nicht-syndromal (THC): 9 Gene (17,7 kb) ANKRD26, CYCS, ETV6, FYB1, IKZF5, MASTL, SRC, TUBB1, WAS Thrombozythämie (THCYT): 3 Gene (6,4 kb) JAK2, MPL, THPO Thrombozytenbedingte Blutungsstörung (BDPLT): 23 Gene (52,8 kb) ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1 Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	4 - 6 Wo	E
<b>Thrombozytopathie (BDPLT, HPS) *</b> Gen-Panel: ID119.01 Thrombozytopathie (BDPLT, HPS): 37 Gene (91,0 kb) ACTN1, ANO6, AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CD36, DTNBP1, EPHB2, FERMT3, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1 Thrombozytenbedingte Blutungsstörung (BDPLT) mit Thrombozytopenie: 15 Gene (37,9 kb) ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14 Thrombozytenbedingte Blutungsstörung (BDPLT) ohne Thrombozytopenie: 10 Gene (20,3 kb) ANO6, EPHB2, GP6, ITGA2B, ITGB3, P2RY12, PTGS1, RASGRP2, TBXA2R, TBXAS1 Hermansky-Pudlak-Syndrom (HPS): 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
<b>Thrombozytopenie (THC) *</b> Gen-Panel: ID104.03 Thrombozytopenie (THC): 49 Gene (98,5 kb) ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CD36, CDC42, CYCS, DIAPH1, ETV6, FLI1, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP9, HOXA11, IKZF1, IKZF5, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, PLAU, PRKACG, PTPRJ, RAP1B, RBM8A, RUNX1, SLFN14, SRC, STIM1, THPO, TPM4, TUBA8, TUBB1, WAS*, WDR1, WIPF1 Thrombozytopenie, nicht-syndromal (THC): 13 Gene (26,3 kb) ANKRD26, CYCS, ETV6, FYB1, GNE, IKZF5, MASTL, PTPRJ, SRC, THPO, TUBA8, TUBB1, WAS Thrombozytenbedingte Blutungsstörung (BDPLT): 16 Gene (38,8 kb) ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14, TPM4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Haut- und Zahnerkrankungen</b>		
<b>Adams-Oliver-Syndrom (AOS) *</b> Gen-Panel: ID259.00, 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 Wo	E
<b>Albinismus, umfassende Diagnostik *</b> 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
<b>Amelogenesis imperfecta (AI) *</b> Gen-Panel: ID232.01, 17 Gene (29,4 kb) ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAHP, RELT, SLC24A4, WDR72	3 - 5 Wo	E
<b>Cowden-Syndrom (CWS) *</b> Gen-Panel: ID075.01, 8 Gene (12,8 kb) AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1	3 - 5 Wo	E
<b>Cutis laxa (ARCL, ADCL) *</b> Gen-Panel: ID109.02, 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 Wo	E
<b>Dyskeratosis congenita (DKC) *</b> Gen-Panel: ID347.01, 15 Gene (24,1 kb) ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53	3 - 5 Wo	E
<b>Ektodermale Dysplasie (ECTD) *</b> 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, IKBK, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LRP6, MSX1, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, WNT10A Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), nicht-syndromal: 20 Gene (27,5 kb) CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBK, 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, IKBK, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2	3 - 5 Wo	E
<b>Ektodermale Dysplasien, umfassende Diagnostik *</b> Gen-Panel: ID366.00 Ektodermale Dysplasien, umfassende Diagnostik: 92 Gene (206,6 kb) ANAPC1, ANTXR1, AP1B1, APCDD1, ARID1A, ARID1B, ATP6V1B2, AXIN2, C3ORF52, CDH1, CDH3, CDSN, CHUK, CST6, CTNND1, CTSK, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, EVC, EVC2, FGF10, FGF2, FGF3, GJA1, GJB2, GJB6, GRHL2, HEPHL1, HOXC13, HR, IFT122, IFT140, IFT43, IFT52, IKBK, 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, RSP04, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SNRPE, SREBF1, ST14, TBC1D24, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, UBR1, WDR19, WDR35, WNT10A Ektodermale Dysplasie (Haare/Zähne/Nägel/Schweißdrüsen), nicht-syndromal: 20 Gene (27,5 kb) CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBK, 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, IKBK, 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, FGF2, FGF3, HEPHL1, IFT122, IFT140, IFT43, IFT52, INSR, KCTD1, KRT14, KRT16, KRT17, KRT81, KRT83, KRT86, NLRP1, PEX1, PEX6, ROGDI, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SREBF1, TBC1D24, UBR1, WDR19, WDR35	4 - 6 Wo	E
<b>Epidermolysis bullosa (EB) *</b> 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, IKBK, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5 Epidermolysis bullosa simplex (EBS): 7 Gene (33,4 kb) CD151, DST, EXPH5, KLHL24, KRT14, KRT5, PLEC Epidermolysis bullosa junctionalis (JEB): 7 Gene (28,4 kb) COL17A1, ITGA3, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2 Epidermolysis bullosa dystrophica (DEB): 1 Gen (8,8 kb) COL7A1 Kindler Epidermolysis bullosa (KNDLRS): 1 Gen (2,0 kb) FERMT1 Syndromale Epidermolysis bullosa: 20 Gene (63,9 kb) ATP2A2, ATP2C1, CAST, CD151, CDSN, CHST8, CSTA, DSG1, DSP, FERMT1, FLG2, IKBK, ITGA3, JUP, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Haut- und Zahnerkrankungen</b>		
<b>Gorlin-Goltz-Syndrom (BCNS) und ähnliche Krankheitsbilder *</b> Gen-Panel: ID174.01, 7 Gene (23,6 kb) CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 Wo	E
<b>Hereditäres Angioödem (HAE) *</b> Gen-Panel: ID345.00, 7 Gene (16,4 kb) ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1	3 - 5 Wo	E
<b>Hermansky-Pudlak-Syndrom (HPS) *</b> Gen-Panel: ID289.00, 11 Gene (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 Wo	E
<b>Hydrops fetalis *</b> 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, CTSB, DHCRT2, DHCRT7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUFB10, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	3 - 5 w	E
<b>Hypotrichose, nicht-syndromale Form (HYPT) *</b> Gen-Panel: ID146.01, 12 Gene (20,1 kb) APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE	3 - 5 Wo	E
<b>Ichthyose *</b> Gen-Panel: ID217.01 Ichthyose: 38 Gene (73,0 kb) ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CLDN10, CSTA, CYP4F22, ELOVL4, ERCC2, FLG, GJB2, KRT1, KRT10, KRT2, LIPN, LORICRIN, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, TGM1 Kongenitale Ichthyose, autosomal-rezessiv (ARCI): 14 Gene (28,7 kb) ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1 Ichthyose, autosomal-dominant und X-chromosomal: 7 Gene (21,3 kb) ASPRV1, FLG, GJB2, KRT1, KRT2, KRT10, STS	3 - 5 Wo	E
<b>Kutanes malignes Melanom (CMM) *</b> 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
<b>Multiples Pterygium-Syndrom *</b> Gen-Panel: ID158.01, 8 Gene (16,8 kb) CHRNA1, CHRN1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	3 - 5 Wo	E
<b>Neurofibromatose (NF) *, #</b> Gen-Panel: ID210.00, 3 Gene (11,6 kb) NF1, NF2, SPRED1	3 - 5 Wo	E
<b>Okulokutaner Albinismus (OCA) *</b> Gen-Panel: ID082.02, 9 Gene (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 Wo	E
<b>Orofaziodigitales Syndrom (OFD) *</b> Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E
<b>Pachyonychia congenita (PC) *</b> Gen-Panel: ID120.01, 13 Gene (30,1 kb) AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3	3 - 5 Wo	E
<b>Palmoplantarkeratose (PPK) *</b> Gen-Panel: ID323.00 Palmoplantarkeratose (PPK): 33 Gene (58,2 kb) AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB3, GJB4, JUP, KDSR, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT10, KRT14, KRT16, KRT17, KRT83, LORICRIN, MBTPS2, PERP, RHBDF2, SERPINB7, SLURP1, SMARCAD1, TAT, TRPM4, TRPV3, WNT10A Palmoplantarkeratose (PPK), nicht-syndromal: 11 Gene (25,7 kb) AAGAB, AQP5, DSG1, DSP, KRT1, KRT6C, KRT9, KRT10, KRT16, SERPINB7, TRPV3 Palmoplantarkeratose (PPK), syndromal: 15 Gene (22,4 kb) CTSC, ENPP1, GJA1, GJB2, KRT14, MBTPS2, JUP, LORICRIN, PERP, RHBDF2, SLURP1, SMARCAD1, TAT, TRPV3, WNT10A Erythrokeratodermia variabilis et progressiva (EKVP): 7 Gene (9,5 kb) GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4 Pachyonychia congenita (PC): 4 Gene (6,1 kb) KRT6A, KRT6B, KRT16, KRT17	3 - 5 Wo	E
<b>Peeling-Skin-Syndrom (PSS) *</b> Gen-Panel: ID309.00, 6 Gene (13,6 kb) CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5	3 - 5 Wo	E
<b>Pierre-Robin-Sequenz *</b> Gen-Panel: ID294.00, 33 Gene (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Haut- und Zahnerkrankungen</b>		
<b>Primäres Lymphödem (LMPHM) *</b> Gen-Panel: ID372.00, 21 Gene (69,4 kb) ADAMTS3, ANGPT2, CALCRL, CCBE1, CELSR1, EPHB4, ERG, FAT4, FLT4, FOXC2, GATA2, GJA1, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, THSD1, TIE1, VEGFC	3 - 5 Wo	E
<b>Progerie und progeroide Syndrome *</b> Gen-Panel: ID147.01, 25 Gene (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 Wo	E
<b>Selektive Zahn-Agenesie (STHAG) *</b> 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
<b>Systemerkrankungen mit multiplen Café-au-lait-Flecken *</b> 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
<b>Tuberöse Sklerose (TSC) *</b> Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
<b>Xeroderma pigmentosum (XP) *</b> Gen-Panel: ID282.00, 10 Gene (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 Wo	E
<b>Herz- und Gefäßerkrankungen</b>		
<b>Alagille-Syndrom (ALGS) *</b> Gen-Panel: ID112.00, 2 Gene (11,1 kb) JAG1, NOTCH2	3 - 5 Wo	E
<b>Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC) *</b> Gen-Panel: ID010.03, 27 Gene (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNT3, TNNT2, TPM1, TTN	4 - 6 Wo	E
<b>Atriumseptumdefekt und Ventrikelseptumdefekt (ASD, VSD, AVSD) *</b> 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
<b>Bikuspide Aortenklappe (AOVD) *</b> Gen-Panel: ID301.00, 6 Gene (16,7 kb) GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2	3 - 5 Wo	E
<b>Bindegewebserkrankungen mit Aortenbeteiligung *</b> Gen-Panel: ID137.05 Bindegewebserkrankungen mit Aortenbeteiligung: 67 Gene (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB3, TGFB3, TGFB3, 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, TGFB3, TGFB3, TGFB3, THSD4 Ehlers-Danlos-Syndrom (EDS): 20 Gene (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan-Syndrom (MFS): 5 Gene (22,2 kb) CBS, FBN1, FBN2, TGFB2, TGFB3 Cutis laxa-Syndrom (ARCL, ADCL): 11 Gene (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTDB1, LTBP4, PYCR1 Stückler-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
<b>Brugada-Syndrom (BRGDA) *</b> 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
<b>CADASIL und CARASIL *, #</b> Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E

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

Erkrankung/Diagnostik	Dauer	Material
<b>Herz- und Gefäßerkrankungen</b>		
<b>Hereditäre hämorrhagische Teleangiektasie (HHT) *</b> Gen-Panel: ID155.01, 7 Gene (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 Wo	E
<b>Herz-Hand-Syndrom *</b> Gen-Panel: ID165.01, 9 Gene (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 Wo	E
<b>Hypertrophe Kardiomyopathie (CMH, HCM) *</b> Gen-Panel: ID007.05, 56 Gene (253,8 kb) ABC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, LAMP2, KRAS, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL	4 - 6 Wo	E
<b>Kardiale Arrhythmien, umfassende Diagnostik *</b> Gen-Panel: ID026.03 Kardiale Arrhythmien, umfassende Diagnostik: 71 Gene (274,2 kb) ABC9, 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) ABC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, 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) ABC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A Familiäres Vorhofflimmern (ATFB): 17 Gene (29,5 kb) ABC9, 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
<b>Kardiomyopathien, umfassende Diagnostik *</b> Gen-Panel: ID027.05 Kardiomyopathien, umfassende Diagnostik: 154 Gene (487,0 kb) ABC9, ACTA1, ACTC1, ACTN2, ALPK3, ANKRD1, APOA1, B2M, BAG3, BAG5, BRAF, CACNA1C, CACNB2, CALR3, CAP2, CASQ2, CAV3, CDH2, COA5, COA6, CORIN, COX15, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DMPK, DNJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGA, FHL1, FHL2, FHOD3, FKRP, FKTN, FLII, FLNC, FTH1, FXN, GAA, GATA4, GATAD1, GET3, GLA, GSN, HADHA, HAMP, HCN4, HFE, HJV, HRAS, ILK, JPH2, JUP, KCNQ1, KIF20A, KLF10, KLHL24, KRAS, KY, LAMA4, LAMP2, LDB3, LIMS2, LMNA, LMOD2, LRRC10, LYZ, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM10, MIB1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOT, MYOZ2, MYPN*, NEBL, NEXN, NKX2-5, NNT, NONO, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, PYROXD1, RAF1, RBM20, RIT1, RPL3L, RRAS2, RYR2, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC40A1, SLC25A4, SOS1, SOS2, SPRED2, SVIL, SYNE1, SYNE2, TAFAZZIN, TBX5, TBX20, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TRPM4, TTN, TTR, UNC45B, VCL, VEZF1 Dilatative Kardiomyopathie (DCM, CMD): 68 Gene (301,9 kb) ABC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1 Hypertrophe Kardiomyopathie (HCM, CMH): 56 Gene (253,8 kb) ABC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL Restriktive Kardiomyopathie (RCM): 15 Gene (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC): 27 Gene (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN Nichtdilatierende linksventrikuläre Kardiomyopathie (NDLVC, LVNC): 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
<b>Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) *</b> Gen-Panel: ID012.03, 9 Gene (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN	3 - 5 Wo	E
<b>Konotrunkale Herzfehlbildung (CTHM) *</b> Gen-Panel: ID160.01, 12 Gene (24,8 kb) CFC1, FLT4, FOXH1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Herz- und Gefäßerkrankungen</b>		
<b>Kongenitale Herzfehler, umfassende Diagnostik *</b> 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, TGFB1, TGFB2, TKT, TLL1, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZFPM2, ZIC3 <b>Isolierte kongenitale Herzfehler: 48 Gene (149,9 kb)</b> 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 <b>Syndromale kongenitale Herzfehler: 109 Gene (354,9 kb)</b> ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFB1, TGFB2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3	4 - 6 Wo	E
<b>Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen *</b> Gen-Panel: ID009.07 Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen: 38 Gene (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 <b>Loeys-Dietz-Syndrom (LDS): 8 Gene (11,1 kb)</b> BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 <b>Nicht-syndromales thorakales Aortenaneurysma (AAT): 11 Gene (24,0 kb)</b> ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 <b>Syndrome mit Aortenaneurysma: 29 Gene (91,7 kb)</b> AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2	4 - 6 Wo	E
<b>Long-QT-Syndrom (LQT) *</b> 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
<b>Marfan-Syndrom (MFS) *, #</b> Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFB1, TGFB2	2 - 4 Wo	E
<b>Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder *</b> Gen-Panel ID194.05 <b>Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder: 48 Gene (153,2 kb)</b> ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, UPF3B, ZDHHC9, ZNF469 <b>Marfan-Syndrom (MFS): 3 Gene (11,8 kb)</b> FBN1, TGFB1, TGFB2 <b>Loeys-Dietz-Syndrom (LDS) und Thorakales Aortenaneurysma (AAT): 17 Gene (42,6 kb)</b> ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 <b>Stickler-Syndrom (STL): 6 Gene (21,1 kb)</b> COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 <b>Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)</b> ADAMTS10, ADAMTS17, FBN1, LTBP2 <b>Ehlers-Danlos-Syndrom (EDS): 10 Gene (39,1 kb)</b> CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	4 - 6 Wo	E
<b>Multipler kongenitaler Herzdefekt (CHTD) *</b> Gen-Panel: ID302.01, 9 Gene (21,5 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, PLXND1, SMAD2, TAB2, ZIC3	3 - 5 Wo	E
<b>Muskelerkrankungen mit Herzbeteiligung *</b> Gen-Panel: ID123.03 <b>Muskelerkrankungen mit Herzbeteiligung: 19 Gene (193,6 kb)</b> BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B <b>Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb)</b> BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B <b>Emery-Dreifuss-Muskeldystrophie (EMDM): 6 Gene (51,8 kb)</b> EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 <b>Duchenne/Becker-Muskeldystrophie (DMD, BMD): 1 Gen (11,1 kb)</b> DMD	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Herz- und Gefäßerkrankungen</b>		
<b>Nichtdilatierende linksventrikuläre Kardiomyopathie (NDLVC, LVNC) *</b> Gen-Panel: ID011.04, 35 Gene (226,1kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 Wo	E
<b>Noonan-Syndrom (NS) *</b> 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
<b>Plötzlicher Herztod *</b> Gen-Panel: ID349.01 <b>Plötzlicher Herztod: 127 Gene (393,9 kb)</b> ABCC9, ACTC1, ACTN2, AKAP9, ALG10B, ALPK3, ANK2, ANKRD1, BAG3, BAG5, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAP2, CASQ2, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGF12, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GJA5, GLA, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLHL24, LAMA4, LAMP2, LDB3, LEMD2, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, NPPA, NUP155, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RANGRF, RBM20, RPL3L, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SEMA3A, SGCD, SLC4A3, SLMAP, SNTA1, TAFAZZIN, TBX5, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSPYL1, TTN, TTR, VCL, VEZF1 <b>Kardiale Arrhythmien (BRGDA, LQT) und plötzlicher Herztod: 74 Gene (281,6 kb)</b> ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FGF12, GJA5, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYBPC3, MYH6, MYL4, NKX2-5, NPPA, NUP155, PPA2, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TSPYL1, TTN <b>Kardiomyopathien (HCM, DCM) und plötzlicher Herztod: 71 Gene (261,3 kb)</b> ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, BAG5, CAP2, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSG2, DSP, EMD, EYA4, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GLA, JPH2, JUP, KLHL24, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, TAFAZZIN, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL, VEZF1	4 - 6 Wo	E
<b>Pulmonale Hypertonie (PAH, PPH) *</b> 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
<b>RASopathien *</b> Gen-Panel: ID015.05 <b>RASopathien: 21 Gene (40,1 kb)</b> BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 <b>Noonan-Syndrom (NS): 15 Gene (24,7 kb)</b> BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 <b>Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb)</b> BRAF, KRAS, MAP2K1, MAP2K2 <b>LEOPARD-Syndrom (LPRD): 3 Gene (6,0 kb)</b> BRAF, PTPN11, RAF1	3 - 5 Wo	E
<b>Restriktive Kardiomyopathie (RCM) *</b> Gen-Panel: ID105.03, 15 Gene (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR	4 - 6 Wo	E
<b>Syndromale kongenitale Herzfehler *</b> Gen-Panel ID252.02 <b>Syndromale kongenitale Herzfehler: 109 Gene (354,9 kb)</b> ABL1, ACTA2, ACTB, CTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFB1, TGFB2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WPCP, YY1AP1, ZEB2, ZIC3 <b>Noonan-Syndrom (NS): 16 Gene (27,4 kb)</b> BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 <b>Kardiofaziokutanes Syndrom (CFC): 4 Gene (5,3 kb)</b> BRAF, KRAS, MAP2K1, MAP2K2 <b>Ritscher-Schinzel-Syndrom (RTSC): 4 Gene (10,0 kb)</b> DPYSL5, CCDC22, VPS35L, WASHC5 <b>Weill-Marchesani-Syndrom (WMS): 4 Gene (20,7 kb)</b> ADAMTS10, ADAMTS17, FBN1, LTBP2 <b>Adams-Oliver-Syndrom (AOS): 6 Gene (23,2 kb)</b> ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ <b>Kabuki-Syndrom (KABUK): 2 Gene (20,8 kb)</b> KDM6A, KMT2D <b>Marfan-Syndrom (MFS): 3 Gene (11,8 kb)</b> FBN1, TGFB1, TGFB2 <b>CHARGE-Syndrom: 2 Gene (11,3 kb)</b> CHD7, SEMA3E <b>VCRL-Syndrom: 3 Gene (4,4 kb)</b> HAAO, KYNU, NADSYN1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Herz- und Gefäßerkrankungen</b>		
<b>Short-QT-Syndrom (SQT) *</b> Gen-Panel: ID233.01, 7 Gene (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3	3 - 5 Wo	E
<b>Sick-Sinus-Syndrom (SSS) *</b> Gen-Panel: ID107.01, 4 Gene (16,5 kb) GNB2, HCN4, MYH6, SCN5A	3 - 5 Wo	E
<b>Speicherkrankheiten mit Herzbeteiligung *</b> Gen-Panel: ID149.03 Speicherkrankheiten mit Herzbeteiligung: 16 Gene (20,7 kb) APOA1, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Kardiale Glykogenspeicherkrankheit (GSD): 3 Gene (5,8 kb) GAA, LAMP2, PRKAG2 Hämochromatose (HFE): 6 Gene (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Amyloidose (AMYLD): 6 Gene (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR	3 - 5 Wo	E
<b>Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD) *</b> 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, TGFB3, TGFB3, TGFB3, THSD4 Nicht-syndromales thorakales Aortenaneurysma (AAT): 10 Gene (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Loeys-Dietz-Syndrom (LDS): 6 Gene (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFB3, TGFB3, TGFB3	3 - 5 Wo	E
<b>Viszerale Heterotaxie (HTX) *</b> Gen-Panel: ID145.01, 18 Gene (72,4 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, DNAAF1, DNAH5, DNAH9, DNAH11, MMP21, MNS1, NODAL, ODAD2, PKD1L1, ZIC3	3 - 5 Wo	E
<b>Zerebrale Kleingefäßerkrankung (BSVD) *</b> Gen-Panel: ID325.00, 3 Gene (12,0 kb) COL4A1, COL4A2, COLGALT1	3 - 5 Wo	E
<b>Zerebrovaskuläre Erkrankungen und Schlaganfall *</b> 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, TGFB3, TGFB3, 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, TGFB3, TGFB3 Moyamoya-Erkrankung (MYMY): 4 Gene (22,2 kb) ACTA2, GUCY1A1, JAG1, RNF213 CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3	4 - 6 Wo	E
<b>HNO-Erkrankungen</b>		
<b>Branchiotorenales Syndrom (BOR) *</b> Gen-Panel: ID315.00, 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 Wo	E
<b>Hypogonadotroper Hypogonadismus mit oder ohne Anosmie (KAL, HH) *</b> 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
<b>Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX) *</b> Gen-Panel: ID290.01, 8 Gene (17,1 kb) AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX, TIMM8A	3 - 5 Wo	E
<b>Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB) *</b> Gen-Panel: ID092.04, 83 Gene (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJKV, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>HNO-Erkrankungen</b>		
<b>Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA) *</b> Gen-Panel: ID091.04, 60 Gene (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1	4 - 6 Wo	E
<b>Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik *</b> Gen-Panel: ID237.03 Nicht-syndromale Schwerhörigkeit, umfassende Diagnostik: 133 Gene (374,8 kb) ABCC1, ACTG1, ADCY1, AFG2B, AIFM1, ATOH1, ATP11A, ATP2B2, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GPRASP2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MAP1B, MARVELD2, MCM2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PI4KB, PJVK, PKHD1L1, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, THOC1, TMC1, TMEM132E, TMIE, TMRSS3, TMTC4, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, USP48, WBP2, WFS1, WHRN Nicht-syndromale Schwerhörigkeit, autosomal-dominant (DFNA): 60 Gene (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24*, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1 Nicht-syndromale Schwerhörigkeit, autosomal-rezessiv (DFNB): 83 Gene (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN Nicht-syndromale Schwerhörigkeit, X-chromosomal (DFNX): 6 Gene (11,8 kb) AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX	4 - 6 Wo	E
<b>Perrault-Syndrom (PRLTS) *</b> Gen-Panel: ID388.00, 9 Gene (14,1 kb) CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, MRPL49, PRORP, TWNK	3 - 5 Wo	E
<b>Syndromale Schwerhörigkeit, umfassende Diagnostik *</b> 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, GSPM2, 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
<b>Usher-Syndrom (USH) *</b> Gen-Panel: ID034.01 Usher-Syndrom (USH): 13 Gene (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN Usher-Syndrom, Typ I (USH1): 6 Gene (26,2 kb) CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G Usher-Syndrom, Typ II (USH2): 4 Gene (40,4 kb) ADGRV1, PDZD7, USH2A, WHRN Usher-Syndrom, Typ III und Typ IV (USH3, USH4): 3 Gene (3,8 kb) ARSG, CLRN1, HARS1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Immunologische Erkrankungen</b>		
<b>Antikörpermangelsyndrome (CVID, AGM) *</b> Gen-Panel: ID382.00 Antikörpermangelsyndrome (CVID, AGM): 38 Gene (70,7 kb) AICDA, BLNK, BTK, CARD11, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, CTNND1, CXCR4, FNIP1, ICOS, IGLL1, IKZF1, IL21, IRF2BP2, LRBA, LRRC8A, MS4A1, NFKB1, NFKB2, OAS1, PIK3CD, PIK3R1, PLCG2, RAC2, SEC61A1, SH3KBP1, SLC39A7, SPI1, TCF3, TNFRSF13B, TNFRSF13C, TOP2B, UNG Allgemeiner variabler Immundefekt (CVID): 14 Gene (28,0 kb) CD19, CD81, CR2, ICOS, IKZF1, IL21, IRF2BP2, LRBA, MS4A1, NFKB1, NFKB2, SEC61A1, TNFRSF13B, TNFRSF13C Agammaglobulinämie (AGM): 11 Gene (16,2 kb) BLNK, BTK, CD79A, CD79B, IGLL1, LRRC8A, PIK3R1, SH3KBP1, SLC39A7, SPI1, TCF3 Immundefekt mit Hyper-IgM (HIGM): 4 Gene (3,2 kb) AICDA, CD40, CD40LG, UNG	3 - 5 Wo	E
<b>Atypisches hämolytisch-urämisches Syndrom (AHUS) *</b> 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
<b>Autoinflammatorische Erkrankungen, umfassende Diagnostik *</b> Gen-Panel: ID087.05 Autoinflammatorische Syndrome, umfassende Diagnostik: 51 Gene (99,3 kb) ADA2, ALPK1, ARPC5, CARD14, DNASE2, DOCK11, DPP9, ELANE, ELF4, HCK, IKBK, IL1RN, IL36RN, JAK1, LPIN2, LYN, MEFV, MVK, NCKAP1L, NFKB1, NLRP4, NLRP1, NLRP12, NLRP3, NOD2, NTRK1, OTULIN, PLCG2, POMP, PSMB4, PSMB8, PSMB9, PSMB10, PSMG2, PSTPIP1, RBCK1, REL, RIPK1, RNF31, SHARPIN, SLC29A3, SOCS1, STING1, SYK, TBK1, TNFAIP3, TNFRSF1A, TNFRSF11A, TRNT1, WDR1 Periodische Fiebersyndrome: 14 Gene (29,9 kb) ELANE, LPIN2, MEFV, MVK, NLRP4, NLRP12, NLRP3, NOD2, OTULIN, PSMB8, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 Wo	E
<b>Chronische Granulomatose (CGD) *</b> Gen-Panel: ID379.00, 8 Gene (8,8 kb) CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2	3 - 5 Wo	E
<b>Hydrops fetalis *</b> Gen-Panel: ID370.00, 148 Gen (404,7 kb) ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCB1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNG, COL2A1, CTSA, DHCR24, DHCR7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHP1L1, 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, RAPS, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	4 - 6 Wo	E
<b>Hyper-IgE-Syndrom mit rekurrenten Infektionen (HIES) *</b> Gen-Panel: ID240.01, 10 Gene (29,5 kb) DOCK8, DSG1, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TYK2, ZNF341	3 - 5 Wo	E
<b>Knochenmarkinsuffizienz (BMF) *</b> Gen-Panel: ID357.00 Knochenmarkinsuffizienz (BMF): 15 Gene (29,9 kb): ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTE1, SRP72, TERC, TERT, TP53, ZCCHC8 Knochenmarkinsuffizienz-Syndrom (BMFS): 8 Gene (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomer-assoziierte Knochenmarkinsuffizienz und/oder Lungenfibrose (PFBMFT): 6 Gene (13,0 kb) PARN, RPA1, RTE1, TERC, TERT, ZCCHC8	3 - 5 Wo	E
<b>Primäre Immundefekte, umfassende Diagnostik *</b> Gen-Panel: ID380.00, 441 Gen (942,4 kb) ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALPI, ALPK1, ANGPT1, ANKZF1, AP1S3, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATAD3A, ATM, ATP6A1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2ORF69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD10, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CBLB, CCB1, CD19, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFHR1, CFI, CFP, CFTR, CHD7, CIB1, CIITA, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNND1, CTSP1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DPP9, DUT, EFL1, ELANE, ELF4, EPG5, ERBIN, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FGL2, FLT3LG, FMNL2, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC3, G6PD, GATA1, GATA2, GF11, GIMAP5, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HPS1, HPS4, HPS6, HSPA1L, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKBK, IKBK, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1R1, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IPO8, IRAK4, IRF1, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, IVNS1ABP, JAGN1, JAK1, JAK3, KCNA5, KDM6A, KMT2A, KMT2D, KRAS, LACC1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYN, LYST, MAGT1, MALT1, MAP3K14, MBL2, MCM10, MCM4, MCTS1, MECOM, MEFV, MOGS, MPEG1, MPO, MRTFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NPC1, NRAS, NSMCE3, NUDCD3, OAS1, ORAI1, OTULIN, PARN, PAX1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLG, PNP, POLA1, POLD1, POLD3, POLE, POLR3A, POLR3C, POLR3F, POMP, PRF1, PRIM1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSTPIP1, PTCRA, PTEN, PTPN2, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RC3H1, RECQL4, REL, RELB, RFX5, RFXANK, RFXAP, RHBDF2, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU7-1, RORC, RPSA, RTE1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCGN, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLC9A3, SLC02A1, SMARCA1, SMARCD2, SNORA31, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STX11, STXBP2, STXBP3, SYK, TAP1, TAP2, TAPBP, TAFAZZIN, TBK1, TBX1, TBX21, TCF3, TCN2, TERC, TERT, TET2, TFR, TGFB1, TGFB2, TICAM1, TIN2, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TOM1, TOP2B, TPP2, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC7A, TYK2, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, XIAP, ZAP70, ZBTB24, ZNF341, ZNF41	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Immunologische Erkrankungen</b>		
<b>Periodische Fiebersyndrome *</b> Gen-Panel: ID088.04, 12 Gene (26,3 kb) ELANE, MEV, MVK, NLR4, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 Wo	E
<b>Neutropenie *</b> Gen-Panel: ID189.02 Neutropenie: 30 Gene (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAZ, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Schwere kongenitale Neutropenie (SCN): 11 Gene (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndrome mit Neutropenie: 21 Gene (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAZ, USB1, VPS13B, WDR1	3 - 5 Wo	E
<b>Schwerer kombinierter Immundefekt (SCID) *</b> Gen-Panel: ID381.00, 21 Gene (47,6 kb) ADA, AK2, BCL11B, CD3E, CD3D, CIITA, CORO1A, DCLRE1C, FOXP1, IL2RG, IL7R, LAT, LIG4, JAK3, NHEJ1, POLD3, PRKDC, PTPRC, RAC2, RAG1, RAG2	3 - 5 Wo	E
<b>Thrombotische Mikroangiopathie (TMA) *</b> Gen-Panel: ID707.00, 23 Gene (44,7 kb) ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD	3 - 5 Wo	E
<b>Lungen- und Respiratorische Erkrankungen</b>		
<b>Interstitielle Lungenerkrankung und Lungenfibrose *</b> 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
<b>Kongenitales zentrales Hypoventilationssyndrom (CCHS) *</b> Gen-Panel: ID185.01, 5 Gene (9,7 kb) LBX1, MECP2, MYO1H, PHOX2B, RET	2 - 4 Wo	E
<b>Lungenkarzinom *</b> 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
<b>Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *</b> Gen-Panel: ID085.03, 50 Gene (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 Wo	E
<b>Pulmonale Hypertonie (PAH, PPH) *</b> 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
<b>Pulmonale Surfactant-Stoffwechselstörung (SMDP) *</b> Gen-Panel: ID168.01, 6 Gene (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 Wo	E
<b>Zystische Fibrose (CF) *</b> Gen-Panel: ID045.00, 1 Gen (4,4 kb) CFTR	2 - 4 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Mikrozephalien und Makrozephalien</b>		
<b>Galloway-Mowat-Syndrom (GAMOS) *</b> Gen-Panel: ID251.01, 10 Gene (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 Wo	E
<b>Hydrozephalie *</b> Gen-Panel: ID221.04 Hydrozephalie: 20 Gene (57,9 kb) AKT3, CCDC88C, CCND2, CFAP43, FANCB, FLVCR2, FOXJ1, GPM2, HYL1, KIF7, L1CAM, MPDZ, P4HB, PIK3R2, POMT1, SEC24D, SMARCC1, TRIM71, WDR81, ZIC3 Kongenitaler Hydrozephalus (HYC): 6 Gene (27,7 kb) CCDC88C, L1CAM, MPDZ, SMARCC1, TRIM71, WDR81 Syndrome mit Hydrozephalus: 14 Gene (29,0 kb) AKT3, CCND2, FANCB, FLVCR2, FOXJ1, GPM2, HYL1, KIF7, L1CAM, P4HB, PIK3R2, POMT1, SEC24D, ZIC3	3 - 5 Wo	E
<b>Intellektuelle Entwicklungsstörung und Makrozephalie *</b> Gen-Panel: ID131.03 Intellektuelle Entwicklungsstörung und Makrozephalie: 48 Gene (147,3 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMC01, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9 Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Makrozephalie: 21 Gene (70,8 kb) CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20 Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Makrozephalie: 10 Gene (20,2 kb) ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMC01, ZBTB7A Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Makrozephalie: 18 Gene (57,9 kb) BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9	4 - 6 Wo	E
<b>Intellektuelle Entwicklungsstörung und Mikrozephalie *</b> Gen-Panel: ID129.02 Intellektuelle Entwicklungsstörung und Mikrozephalie: 80 Gene (229,8 kb) ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNBN1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCF1, HIVEP2, HNMT, HNRNP2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2 Intellektuelle Entwicklungsstörung, autosomal-dominant (MRD) und Mikrozephalie: 32 Gene (100,5 kb) AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNBN1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2 Intellektuelle Entwicklungsstörung, autosomal-rezessiv (MRT) und Mikrozephalie: 21 Gene (41,0 kb) ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73 Intellektuelle Entwicklungsstörung, X-chromosomal (MRX) und Mikrozephalie: 27 Gene (88,3 kb) ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCF1, HNRNP2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2	4 - 6 Wo	E
<b>Mikrozephalie, umfassende Diagnostik *</b> Gen-Panel: ID069.02 Mikrozephalie, umfassende Diagnostik: 129 Gene (371,7 kb) ADARB1, ANKLE2, ARCN1, ARFGF2, ASPM, ATR, BLM, CARS1, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CENPT, CEP135, CEP152, CEP63, CIT, CKAP2L, COPB1, COPB2, CRIPT, CTNBN1, CTU2, DIAPH1, DNA2, DNMT3A, DONSON, DPP6, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, ERCC1, ERCC2, ERCC5, ERCC6, EXOC8, EXT2, FOXG1, GEMIN4, GPT2, IER3IP1, KAT6A, KATNB1, KCNA4, KIF11, KIF14, KNL1, LAGE3, LMNB1, LMNB2, MCPH1, MED17, MFSD2A, 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, RITN, SARS1, SASS6, SLC1A4, SLC25A19, SLC9A6, SMPD4, SPOP, STAG2, STAMBP, STIL, SVBP, TBC1D20, THOC6, TMX2, TOP3A, TP53RK, TPRKB, TRAP1, 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, TRAP1 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 Zerebrokulofazioskelettales Syndrom (COFS): 4 Gene (11,3 kb) ERCC1, ERCC2, ERCC5, ERCC6 Mikrozephalie und Chorioretinopathie (MCCRP): 4 Gene (13,5 kb) PLK4, TUBGCP4, TUBGCP6, KIF11 Neurologische Entwicklungsstörung mit Mikrozephalie (NEDM): 21 Gene (39,7 kb) ADARB1, COPB1, CTNBN1, DYNC1I2, EXOC8, GEMIN4, GPT2, MFSD2A, MTHFS, NARS1, PLAA, PRUNE1, PUS3, SPOP, STAG2, TMX2, TRAPPC6B, SARS1, SMPD4, SVBP, VARS1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Mikrozephalien und Makrozephalien</b>		
<b>Makrozephalie, umfassende Diagnostik *</b> Gen-Panel: ID070.03 Makrozephalie, umfassende Diagnostik: 135 Gene (364,6 kb) ABCG9, ADK, AKT1, AKT3, ALKBH8, AMER1, ANKH, APC2, ASPA, ASXL2, BRAF, BRWD3, CAMK2G, CCDC22, CCDC88C, CCND2, CDH2, CDKN1C, CHD1, CHD3, CHD8, CRADD, CSF1R, CUL4B, DIS3L2, DDX3X, DEAF1, DNMT3A, DVL1, DVL3, EED, EML1, EXT2, EZH2, FGFR3, FIBP, FMR1, FOXP1, GATAD2B, GCDH, GFAP, GLI3, GPC3, GPC4, GRIA3, GUSB, H1-4, HEPACAM, HERC1, HRAS, HSD17B4, HUWE1, IGBP1, INPPL1, KDM5C, KIF7, KPTN, KRAS, L1CAM, LAMB1, LBR, LZTR1, MAN2B1, MAP2K1, MAP2K2, MAPK1, MECP2, MED12, MITF, MLC1, MPDZ, MRAS, MSL3, MTOR, NF1, NFIB, NFIX, NONO, NRAS, NSD1, NXN, OFD1, OPHN1, PAK1, PHF21A, PIGA, PIGM, PIGN, PIGT, PIK3CA, PIK3R2, PKDCC, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, PTPN11, RAB39B, RAC1, RAF1, RIN2, RIT1, RNF125, RNF135, ROR2, RRAS2, SEC23B, SETD2, SHANK3, SHOC2, SHROOM4, SNX10, SOS1, SOS2, SPOP, SPRED1, SPRED2, STRADA, SUFU, SUZ12, TBC1D7, TCIRG1, TMC01, TNFRSF11A, TRIO, TRIP12, UPF3B, WASHC5, WDR81, WNT5A, ZBTB7A, ZBTB20, ZBTB42, ZDHHC9 Cowden-Syndrom (CWS): 4 Gene (8,2kb) AKT1, PIK3CA, PTEN, SEC23 Robinow-Syndrom (RRS, DRS): 5 Gene (9,5 kb) DVL1, DVL3, NXN, ROR2, WNT5A Großwuchssyndrome: 15 Gene (38,9 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, FIBP, GPC3, GPC4, NFIX, NSD1, OFD1, PTEN, RNF125, SETD2, SUZ12 RASopathien: 21 Gene (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Intellektuelle Entwicklungsstörungen und Makrozephalie: 49 Gene (147,9 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, H1-4, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMC01, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9	4 - 6 Wo	E
<b>Pontozebelläre Hypoplasie (PCH) *</b> 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
<b>Primäre Mikrozephalie, autosomal-rezessiv (MCPH) *</b> 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
<b>Seckel-Syndrom (SCKL) *</b> Gen-Panel: ID113.00, 9 Gene (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAP	3 - 5 Wo	E
<b>Wachstumsstörung und Makrozephalie *</b> 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
<b>Mitochondriopathien</b>		
<b>Coenzym-Q10-Mangel (COQ10D) *</b> 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
<b>Defizienz der mitochondrialen Komplexe I bis V (MCDN) *</b> Panel: ID074.03 Defizienz der mitochondrialen Komplexe I bis V (MCDN): 83 Gene (60,7 kb) ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX5A, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, CYC1, DNAJC30, FOXRED1, LRPPRC, LYRM7*, MTFMT, NDUFA1, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, PET100, PET117, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SURF1, TACO1, TIMMDC1, TMEM70, TMEM126B, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ Mitochondrialer Komplex-I-Mangel, kernkodierter Typ (MC1DN): 39 Gene (29,3 kb) ACAD9, DNAJC30, FOXRED1, MTFMT, NDUFA1, NDUFA2, NDUFA6, NDUFA8, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB7, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, TIMMDC1, TMEM126B Mitochondrialer Komplex-II-Mangel, kernkodierter Typ (MC2DN): 4 Gene (3,7 kb) SDHA, SDHAF, SDHB, SDHD Mitochondrialer Komplex-III-Mangel, kernkodierter Typ (MC3DN): 10 Gene (7,1 kb) BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1 Mitochondrialer Komplex-IV-Mangel, kernkodierter Typ (MC4DN): 23 Gene (15,8 kb) COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX11, COX14, COX15, COX16, COX20, COX5A, LRPPRC, NDUFA4, PET100, PET117, SCO1, SCO2, SURF1, TACO1 Mitochondrialer Komplex-V-(ATP-Synthase)-Mangel, kernkodierter Typ (MC5DN): 7 Gene (4,8 kb) ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATPAF2, TMEM70	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Mitochondriopathien</b>		
<b>Kombinierter Defekt der oxidativen Phosphorylierung (COXPD) *</b> Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCO3, RMND1, SFXN4, SLC25A26, TARS2, TMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 Wo	E
<b>Lebersche hereditäre Optikusneuropathie (LHON) #</b> 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
<b>MELAS-Syndrom #</b> 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
<b>Mitochondriales DNA-Depletionssyndrom (MTDPS) *</b> Gen-Panel: ID324.00, 19 Gene (25,6 kb) AGK, DGUOK, FBXL4, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUGL1, TFAM, TK2, TWNK, TYMP	3 - 5 Wo	E
<b>Mitochondriale Myopathie / Enzephalopathie #</b> Gen-Panel: ID702.00, 27 Gene (7,0 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND2, MT-ND5, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TW	3 - 5 Wo	E
<b>Mitochondriengenom #</b> Gen-Panel: ID703.00, 37 Gene (11,4 kb) MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY	3 - 5 Wo	E
<b>Nukleär-kodierte mitochondriale Erkrankungen *</b> 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, BTBD, C1QBP, CARS2, CISD2, COA3, COA5, COA6, COA8, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX5A, COX6A2, COX6B1, CPS1, CPT1A, CPT2, CYB5R3, CYC1, CYCS, CYP27A1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAC19, 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, MIEP, MLYCD, MMAA, MMAB, MMADHC, MMUT, MPV17, MRM2, MRPL12, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS28, MRPS34, MRPS7, MTFMT, MTO1, MTPAP, MTRFR, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NSUN3, NUBPL, OAT, OPA1, OPA3, OTC, OXCT1, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PET117, PINK1, PNKD, PNPLA2, PNPT1, POLG, POLG2, PPOX, PRKN, PTCO3, 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, SUGL1, SURF1, TACO1, TFAZZIN, TARS2, TFAM, TIMM22, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPX1, TRIT1, TRMT10C, TRMT5, TRMU, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UNG, UQC2, UQC3, UQCRB, UQCRC2, UQCRC3, UQCRC4, UQCRC5, UQCRC6, UQCRC7, UQCRC8, UQCRC9, UQCRC10, UQCRC11, UQCRC12, UQCRC13, UQCRC14, UQCRC15, UQCRC16, UQCRC17, UQCRC18, UQCRC19, UQCRC20, UQCRC21, UQCRC22, UQCRC23, UQCRC24, UQCRC25, UQCRC26, UQCRC27, UQCRC28, UQCRC29, UQCRC30, UQCRC31, UQCRC32, UQCRC33, UQCRC34, UQCRC35, UQCRC36, UQCRC37, UQCRC38, UQCRC39, UQCRC40, UQCRC41, UQCRC42, UQCRC43, UQCRC44, UQCRC45, UQCRC46, UQCRC47, UQCRC48, UQCRC49, UQCRC50, UQCRC51, UQCRC52, UQCRC53, UQCRC54, UQCRC55, UQCRC56, UQCRC57, UQCRC58, UQCRC59, UQCRC60, UQCRC61, UQCRC62, UQCRC63, UQCRC64, UQCRC65, UQCRC66, UQCRC67, UQCRC68, UQCRC69, UQCRC70, UQCRC71, UQCRC72, UQCRC73, UQCRC74, UQCRC75, UQCRC76, UQCRC77, UQCRC78, UQCRC79, UQCRC80, UQCRC81, UQCRC82, UQCRC83, 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UQCRC1077, UQCRC1078, UQCRC1079, UQCRC1080, UQCRC1081, UQCRC1082, UQCRC1083, UQCRC1084, UQCRC1085, UQCRC1086, UQCRC1087, UQCRC1088, UQCRC1089, UQCRC1090, UQCRC1091, UQCRC1092, UQCRC1093, UQCRC1094, UQCRC1095, UQCRC1096, UQCRC1097, UQCRC1098, UQCRC1099, UQCRC1100, UQCRC1101, UQCRC1102, UQCRC1103, UQCRC1104, UQCRC1105, UQCRC1106, UQCRC1107, UQCRC1108, UQCRC1109, UQCRC1110, UQCRC1111, UQCRC1112, UQCRC1113, UQCRC1114, UQCRC1115, UQCRC1116, UQCRC1117, UQCRC1118, UQCRC1119, UQCRC1120, UQCRC1121, UQCRC1122, UQCRC1123, UQCRC1124, UQCRC1125, UQCRC1126, UQCRC1127, UQCRC1128, UQCRC1129, UQCRC1130, UQCRC1131, UQCRC1132, UQCRC1133, UQCRC1134, UQCRC1135, UQCRC1136, UQCRC1137, UQCRC1138, UQCRC1139, UQCRC1140, UQCRC1141, UQCRC1142, UQCRC1143, UQCRC1144, UQCRC1145, UQCRC1146, UQCRC1147, UQCRC1148, UQCRC1149, UQCRC1150, UQCRC1151, UQCRC1152, UQCRC1153, UQCRC1154, UQCRC1155, UQCRC1156, UQCRC1157, UQCRC1158, UQCRC1159, UQCRC1160, UQCRC1161, UQCRC1162, UQCRC1163, UQCRC1164, UQCRC1165, UQCRC1166, UQCRC1167, UQCRC1168, UQCRC1169, UQCRC1170, UQCRC1171, UQCRC1172, UQCRC1173, UQCRC1174, UQCRC1175, UQCRC1176, UQCRC1177, UQCRC1178, UQCRC1179, UQCRC1180, UQCRC1181, UQCRC1182, UQCRC1183, UQCRC1184, UQCRC1185, UQCRC1186, UQCRC1187, UQCRC1188, UQCRC1189, UQCRC1190, UQCRC1191, UQCRC1192, UQCRC1193, UQCRC1194, UQCRC1195, UQCRC1196, UQCRC1197, UQCRC1198, UQCRC1199, UQCRC1200, UQCRC1201, UQCRC1202, UQCRC1203, UQCRC1204, UQCRC1205, UQCRC1206, UQCRC1207, UQCRC1208, UQCRC1209, UQCRC1210, UQCRC1211, UQCRC1212, UQCRC1213, UQCRC1214, UQCRC1215, UQCRC1216, UQCRC1217, UQCRC1218, UQCRC1219, UQCRC1220, UQCRC1221, UQCRC1222, UQCRC1223, UQCRC1224, UQCRC1225, UQCRC1226, UQCRC1227, UQCRC1228, UQCRC1229, UQCRC1230, UQCRC1231, UQCRC1232, UQCRC1233, UQCRC1234, UQCRC1235, UQCRC1236, UQCRC1237, UQCRC1238, UQCRC1239, UQCRC1240, UQCRC1241, UQCRC1242, UQCRC1243, UQCRC1244, UQCRC1245, UQCRC1246, UQCRC1247, UQCRC1248, UQCRC1249, UQCRC1250, UQCRC1251, UQCRC1252, UQCRC1253, UQCRC1254, UQCRC1255, UQCRC1256, UQCRC1257, UQCRC1258, UQCRC1259, UQCRC1260, UQCRC1261, UQCRC1262, UQCRC1263, UQCRC1264, UQCRC1265, UQCRC1266, UQCRC1267, UQCRC1268, UQCRC1269, UQCRC1270, UQCRC1271, UQCRC1272, UQCRC1273, UQCRC1274, UQCRC1275, UQCRC1276, UQCRC1277, UQCRC1278, UQCRC1279, UQCRC1280, UQCRC1281, UQCRC1282, UQCRC1283, UQCRC1284, UQCRC1285, UQCRC1286, UQCRC1287, UQCRC1288, UQCRC1289, UQCRC1290, UQCRC1291, UQCRC1292, UQCRC1293, UQCRC1294, UQCRC1295, UQCRC1296, UQCRC1297, UQCRC1298, UQCRC1299, UQCRC1300, UQCRC1301, UQCRC1302, UQCRC1303, UQCRC1304, UQCRC1305, UQCRC1306, UQCRC1307, UQCRC1308, UQCRC1309, UQCRC1310, UQCRC1311, UQCRC1312, UQCRC1313, UQCRC1314, UQCRC1315, UQCRC1316, UQCRC1317, UQCRC1318, UQCRC1319, UQCRC1320, UQCRC1321, UQCRC1322, UQCRC1323, UQCRC1324, UQCRC1325, UQCRC1326, UQCRC1327, UQCRC1328, UQCRC1329, UQCRC1330, UQCRC1331, UQCRC1332, UQCRC1333, UQCRC1334, UQCRC1335, UQCRC1336,		

Erkrankung/Diagnostik	Dauer	Material
<b>Multisystem-Fehlbildungssyndrome</b>		
<b>Cornelia-de-Lange-Syndrom (CDLS) *</b> Gen-Panel: ID033.02, 8 Gene (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 Wo	E
<b>Fehlbildungssyndrome mit überwiegend fazialer Beteiligung *</b> Gen-Panel: ID279.00 Fehlbildungssyndrome mit überwiegend fazialer Beteiligung: 25 Gene (90,9 kb) C2CD3, COL11A2, CPLANE1, DDX59, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GRIP1, IFT57, INTU, KIAA0753, MEGF8, MYH3, MYMK, MYT1, OFD1, RAB23, RBM10, TCTN3, TGDS, TMEM107, TNNT2, TWIST1 Akrozephalosyndaktylie-Syndrom (ACS): 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107 Fraser-Syndrom (FRASRS): 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1 Pierre-Robin-Syndrom: 4 Gene (9,7 kb) COL11A2, MYMK, RBM10, TGDS	3 - 5 Wo	E
<b>FG-Syndrom (FGS) *</b> Gen-Panel: ID215.00, 3 Gene (17,2 kb) CASK, FLNA, MED12	3 - 5 Wo	E
<b>Großwuchssyndrome (SOTOS, BWS) *</b> Gen-Panel: ID073.05 Großwuchssyndrome (SOTOS, BWS): 15 Gene (41,2 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, PDGFRB, PTEN, SETD2, SUZ12, RNF125 Sotos-ähnliche Großwuchssyndrome: 6 Gene (23,1 kb) EED, EZH2, NFIX, NSD1, SETD2, SUZ12 Beckwith-Wiedemann-ähnliche Großwuchssyndrome: 5 Gene (10,1 kb) CDKN1C, DIS3L2, GPC3, GPC4, OFD1	3 - 5 Wo	E
<b>Kabuki-Syndrom (KABUK) *, #</b> Gen-Panel: ID127.00, 2 Gene (20,8 kb) KDM6A, KMT2D	3 - 5 Wo	E
<b>Marfan-Syndrom (MFS) *, #</b> Gen-Panel: ID022.00, 3 Gene (11,8 kb) FBN1, TGFBFR1, TGFBFR2	2 - 4 Wo	E
<b>Noonan-Syndrom (NS) *</b> 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
<b>Orofaziodigitales Syndrom (OFD) *</b> Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E
<b>RASopathien *</b> 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 Kardiofaziokutanen 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
<b>Rubinstein-Taybi-Syndrom (RSTS) *</b> Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
<b>Stickler-Syndrom (STL) *</b> Gen-Panel: ID062.00, 6 Gene (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 Wo	E
<b>Tuberöse Sklerose (TSC) *</b> Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
<b>VACTERL-Assoziation *</b> Gen-Panel: ID258.02, 27 Gene (72,8 kb) BRCA2, CHD7, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGF8, FOXF1, GLI3, HAAO, HOXD13, HSPA6, MNX1, RECQL4, SALL1, KYNU, MYCN, NADSYN1, RAD51C, TRAP1, WBP11, ZIC3	3 - 5 Wo	E
<b>Weill-Marchesani-Syndrom (WMS) *</b> Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
<b>Zellweger-Syndrom (ZWS) *</b> Gen-Panel: ID084.00, 14 Gene (22,3 kb) ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	3 - 5 Wo	E



Erkrankung/Diagnostik	Dauer	Material
<b>Muskelerkrankungen</b>		
<b>Distale Arthrogrypose (DA) *</b> Gen-Panel: ID196.02, 11 Gene (40,5 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNT2, TNNT3, TPM2, UBA1	3 - 5 Wo	E
<b>Distale hereditäre motorische Neuronopathie (HMND, HMNR) *</b> Gen-Panel: ID254.01 Distale hereditäre motorische Neuronopathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Distale motorische Neuronopathie, autosomal-dominant (HMND): 15 Gene (38,4 kb) BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1 Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 10 Gene (14,4 kb) ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1	3 - 5 Wo	E
<b>Distale Myopathie (MPD) *</b> Gen-Panel: ID328.01, 30 Gene (186,0 kb) ACTA1, ACTN2, ADSS1, ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, HNRNPA1, HNRPA2B1, HSPB8, KY, LDB3, MATR3, MYH7, MYOT, NEB, PYROXD1, SMPX, SQSTM1, TIA1, TCAP, TTN, VCP	4 - 6 Wo	E
<b>Emery-Dreifuss-Muskeldystrophie (EDMD) *</b> Gen-Panel: ID121.00, 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43	3 - 5 Wo	E
<b>Gluedergürtelmuskeldystrophie (LGMD) *</b> Gen-Panel: ID122.04 Gluedergürtelmuskeldystrophie (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 Gluedergürtelmuskeldystrophie, 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 Gluedergürtelmuskeldystrophie, autosomal-dominant (LGMDD): 7 Gene (23,2 kb) CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3 Gluedergürtelmuskeldystrophie-Dystroglykanopathie (MDDGC): 11 Gene (17,6 kb) CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2 Ullrich-Muskeldystrophie (UCMD): 3 Gene (15,7 kb) COL6A1, COL6A2, COL6A3 Bethlem-Myopathie (BTHLM): 3 Gene (15,7 kb) COL6A1, COL6A2, COL6A3	4 - 6 Wo	E
<b>Kongenitales myasthenes Syndrom (CMS) *</b> Gen-Panel: ID130.00 Kongenitales myasthenes Syndrom (CMS): 25 Gene (55,9 kb) AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, MYO9A, PREPL, RAPSN, SCN4A, SLC5A7, SLC18A3, SLC25A1, SNAP25, SYT2, VAMP1 Kongenitales myasthenes Syndrom (CMS), präsynaptisch: 8 Gene (21,9 kb) AGRN, CHAT, MYO9A, SLC5A7, SLC18A3, SLC25A1, SYT2, VAMP1 Kongenitales myasthenes Syndrom (CMS), synaptisch oder postsynaptisch: 18 Gene (40,1 kb) AGRN, ALG2, ALG14, CHRNA1, CHRNB1, CHRND, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, PREPL, RAPSN, SCN4A, SNAP25	3 - 5 Wo	E
<b>Kongenitale Myopathie (CMYP) *</b> Gen-Panel: ID212.02 Kongenitale Myopathie (CMYP): 50 Gene (269,7 kb) ACTA1, ACTN2, BAG3, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, CRYAB, DES, DNAJB4, DNM2, FLNC, FXR1, HACD1, KBTBD13, KLHL40, KLHL41, KY, LDB3, LMOD2, MAP3K20, MEGF10, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYL2, MYOD1, MYOT, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SVIL, TPM2, SELENON, SPEG, STAC3, TNNC2, TNNT1, TPM2, TPM3, TTN, UNC45B Central-Core-Myopathie (CCD): 1 Gen (15,1 kb) RYR1 Nemalin-Myopathie (NEM): 11 Gene (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3 Zentronukleäre Myopathie (CNM): 7 Gene (20,3 kb) BIN1, CCDC78, DNM2, MAP3K20, MTM1, MTMR14, SPEG Myofibrilläre Myopathie (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYOT, PYROXD1, TTN	4 - 6 Wo	E
<b>Letales kongenitales Kontraktursyndrom (LCCS) *</b> 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
<b>Muskeldystrophie-Dystroglykanopathie (MDDG) *</b> 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

Erkrankung/Diagnostik	Dauer	Material
<b>Muskelerkrankungen</b>		
<b>Muskeldystrophie, Typ Duchenne oder Becker (DMD, BMD) *</b> Gen-Panel: ID256.00, 1 Gen (11,1 kb) DMD	2 - 4 Wo	E
<b>Muskelerkrankungen mit Herzbeteiligung *</b> Gen-Panel: ID123.03 Muskelerkrankungen mit Herzbeteiligung: 19 Gene (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrilläre Muskelerkrankung (MFM): 12 Gene (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss-Muskeldystrophie (EMDM): 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker-Muskeldystrophie (DMD, BMD): 1 Gen (11,1 kb) DMD	4 - 6 Wo	E
<b>Muskelerkrankungen, umfassende Diagnostik *</b> 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, CHRN1, CHRN2, CHRNE, CHRN3, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DGU0K, 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, GMPBB, 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, OPA1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSIN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SLC25A3, SLC25A4, SLC25A7, SMCHD1, SMN1, SNAP25, SPEG, SPG7, SPG11, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TCAP, TFAM, TIA1, TK2, TMEM43, TNNT1, TNNT2, 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, TNNT2, 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, GMPBB, HNRNPDL, LAMA2, LARGE1, LIMS2, LMNA, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RXYLT1, SGCA, SGCB, SGC, SGC, SGC, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	4 - 6 Wo	E
<b>Myofibrilläre Myopathie (MFM) *</b> 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
<b>Myotonie *</b> Gen-Panel: ID255.00, 5 Gene (25,0 kb) ATP2A1, CLCN1, HINT1, HSPG2, SCN4A	3 - 5 Wo	E
<b>Nemalin-Myopathie (NEM) *</b> Gen-Panel: ID199.00, 11 Gene (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3	3 - 5 Wo	E
<b>Periodische Paralyse *</b> Gen-Panel: ID253.00, 7 Gene (16,6 kb) CACNA1S, KCNE3, KCNJ2, KCNJ5, KCNJ12, KCNJ18, SCN4A	3 - 5 Wo	E
<b>Spinale Muskelatrophie (SMA) *</b> Gen-Panel: ID152.02 Spinale Muskelatrophie (SMA): 39 Gene (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1 Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB Distale spinale Muskelatrophie (DSMA, HMN): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	3 - 5 Wo	E
<b>Walker-Warburg-Syndrom (WWS, MDDGA) *</b> Gen-Panel: ID178.00, 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPBB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wo	E
<b>Zentronukleäre Myopathie (CNM) *</b> Gen-Panel: ID257.00, 7 Gene (21,4 kb) CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Neurodegenerative Erkrankungen</b>		
<b>Alzheimer-Krankheit (AD) *</b> Gen-Panel: ID157.01, 7 Gene (16,8 kb) ABCA7, ADAM10, APOE, APP, PSEN1, PSEN2, TF	3 - 5 Wo	E
<b>Amyotrophe Lateralsklerose (ALS) *</b> Gen-Panel: ID209.03 Amyotrophe Lateralsklerose (ALS): 35 Gene (80,2 kb) ALS2, ANG, ANXA11, ATXN2, C9ORF72, CCFN, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, LRP12, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1*, SPG11, SPTLC1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP Amyotrophe Lateralsklerose (ALS), juvenile Form: 5 Gene (22,4 kb) ALS2, SETX, SIGMAR1, SPG11, SPTLC1 Amyotrophe Lateralsklerose und Frontotemporale Demenz (ALS, FTDALS): 14 Gene (22,6 kb) C9ORF72, CCFN, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP	3 - 5 Wo	E
<b>Basalganglien-Kalzifikation (IBGC) *</b> Gen-Panel: ID327.00, 6 Gene (11,2 kb) JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1	3 - 5 Wo	E
<b>Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN) *</b> Gen-Panel: ID052.03 Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT2, HMSN): 45 Gene (111,8 kb) AARS1, AIFM1, ATP1A1, CADM3, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, JAG1, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PNKP, PRPS1, RAB7A, SLC12A6, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth-Neuropathie, axonal, dominant (CMT2): 26 Gene (74,2 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, JAG1, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, SLC12A6, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, axonal, rezessiv (CMT2): 10 Gene (24,4 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MFN2, MME, MPV17, PNKP, SPG11, TRIM2 Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Charcot-Marie-Tooth-Neuropathie mit Optikusatrophy (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46	3 - 5 Wo	E
<b>Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN) *</b> Gen-Panel: ID051.03 Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT1, CMT4, HMSN): 29 Gene (69,5 kb) CNTNAP1, COX6A1, DNM2, EGR2, FBLN5, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, ITPR3, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, dominant (CMT1): 11 Gene (19,0 kb) EGR2, FBLN5, GDAP1, ITPR3, LITAF, MPZ, NEFL, POLR3B, PMP2, PMP22 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-rezessiv (CMT4): 12 Gene (33,8 kb) EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Hypertrophe Dejerine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb) EGR2, MPZ, CNTNAP1	3 - 5 Wo	E
<b>Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik *</b> Gen-Panel: ID312.02 Charcot-Marie-Tooth-Neuropathie (CMT, HMSN), umfassende Diagnostik: 87 Gene (217,3 kb) AARS1, AIFM1, ARHGEF10, ATP1A1, ATP7A, BSCL2, CADM3, CNTNAP1, COQ7, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SORD, SPG11, SPTAN1, SPTLC1, SURF1, SYT2, TRIM2, TRPV4, VCP, VRK1, VWA1, WARS1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2): 35 Gene (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophe Déjerine-Sottas-Neuropathie (CMT3, DSS): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Charcot-Marie-Tooth-Neuropathie mit Optikusatrophy (CMT6, HMSN6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46 Distale motorische Neuropathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Neurodegenerative Erkrankungen</b>		
<b>CADASIL und CARASIL * , #</b> Gen-Panel: ID167.01, 3 Gene (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 Wo	E
<b>Choreatiforme Bewegungsstörungen *</b> 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
<b>Distale hereditäre motorische Neuronopathie (HMND, HMNR) *</b> Gen-Panel: ID254.01 Distale hereditäre motorische Neuronopathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Distale motorische Neuronopathie, autosomal-dominant (HMND): 15 Gene (38,4 kb) BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1 Distale motorische Neuronopathie, autosomal-rezessiv (HMNR): 10 Gene (14,4 kb) ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1	3 - 5 Wo	E
<b>Dystonie (DYT) *</b> Gen-Panel: ID128.04 Dystonie (DYT): 25 Gene (55,9 kb) ACTB, ADCY5, ANO3, AOEPEP, ATP1A3, COL6A3, ECHS1, GCH1, GNAL, HPCA, KCTD17, KMT2B, MECP, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VPS16 Dystonie (DYT), isolierte Form: 15 Gene (39,4 kb) ADCY5, ANO3, COL6A3, ECHS1, GNAL, HPCA, KCNN2, KCTD17, KMT2B, SHQ1, SLC2A1, THAP1, TOR1A, TUBB4A, VPS11 Dystonie (DYT), kombinierte Form: 29 Gene (47,2 kb) ACTB, ADCY5, AOEPEP, ATP1A3, ATP5MC3, BCAP31, ECHS1, EIF2AK2, GCH1, MECP, NUP54, PLA2G6, PNKD, PRKRA, PRRT2, SGCE, SHQ1, SLC2A1, SLC6A3, SLC18A2, SLC30A10, SLC39A14, SPR, TAF1, TH, TIMM8A, TMEM151A, VPS16, WARS2	3 - 5 Wo	E
<b>Episodische Ataxie (EA) *</b> Gen-Panel: ID184.02, 6 Gene (20,6 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3	3 - 5 Wo	E
<b>Episodisches Schmerzsyndrom (FEPS) *</b> Gen-Panel: ID268.00, 4 Gene (20,5 kb) SCN9A, SCN10A, SCN11A, TRPA1	3 - 5 Wo	E
<b>Essentieller Tremor (ETM) *</b> Gen-Panel: ID195.01, 4 Gene (16,6 kb) DRD3, FUS, SCN4A, TENM4	3 - 5 Wo	E
<b>Frontotemporale Demenz (FTD) *</b> Gen-Panel: ID310.01 Frontotemporale Demenz (FTD): 19 Gene (29,3 kb) C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporale Demenz und/oder Amyotrophe Lateralsklerose (FTD, FTDALS): 17 Gene (27,1 kb) C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporale Demenz, Einschlusskörpermyopathie und Paget-Syndrom (IBMPFD): 3 Gene (4,6 kb) HNRNPA1, HNRNPA2B1, VCP	3 - 5 Wo	E
<b>Hereditäre Ataxien, umfassende Diagnostik *</b> Gen-Panel: ID276.03 Hereditäre Ataxien, umfassende Diagnostik: 128 Gene (373,1 kb) ABCB7, ABHD12, ACO2, AFG3L2, ANGPL1, AIFM1, ANO10, APOB, APTX, ATCAY, ATG5, ATG7, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CASK, CCDC88C, CHP1, CLCN2, CLN5, COA7, COQ2, COQ4, COQ8A, CWF19L1, CYP27A1, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, FLVCR1, GDAP2, GOSR2, GRID2, GRM1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, LAMA1, MARS2, MME, MRE11, MTCL1, MTPAP, MTPP, NBN, NKX6-2, NPTX1, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PITRM1, PLD3, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRDX3, PRKCG, PRPS1, PTF1A, PUM1, RFC1, RNF216, RNU12, RUBCN, SACS, SAMD9L, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC2A1, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VPS41, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1 Episodische Ataxie (EA): 6 Gene (20,5 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3 Spastische Ataxie (SPAX): 7 Gene (25,6 kb) AFG3L2, CHP1, COQ4, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1 Spinocerebelläre Ataxie, autosomal-dominant (SCA): 27 Gene (91,6 kb) AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPTX1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2 Spinocerebelläre Ataxie, autosomal-rezessiv (SCAR, SCAN): 32 Gene (106,7 kb) ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, UBA5, VPS13D, VPS41, VWA3B, WWOX, XRCC1 Zerebelläre Ataxie mit mentaler Retardierung (CAMRQ): 4 Gene (12,9 kb) ATP8A2, CA8, VLDLR, WDR81 Zerebelläre Ataxie, X-chromosomal: 7 Gene (15,9 kb) ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6 Ataxie mit okulomotorische Apraxie (AOA): 4 Gene (13,3 kb) APTX, PIK3R5, PNKP, SETX Ataxia teleangiectatica (AT): 5 Gene (15,4 kb) APTX, ATM, MRE11, NBN, PCNA	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Neurodegenerative Erkrankungen</b>		
<b>Hereditäre Neuropathien, umfassende Diagnostik *</b> Gen-Panel: ID374.00 Hereditäre Neuropathien, umfassende Diagnostik: 134 Gene (343,6 kb) AARS1, ABCA1, ACOX1, AIFM1, AR, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BICD2, BSCL2, CADM3, CFAP276, CHCHD10, CNTNAP1, COQ7, COX20, COX6A1, CPOX, CYP27A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, GSN, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MAG, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PDXK, PIGB, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PPOX, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN9A, SCN10A, SCN11A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A6, SLC5A7, SLC5A2, SLC52A3, SMN1, SORD, SPG11, SPTAN1, SPTLC1, SPTLC2, SURF1, SYT2, TECPR2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VCP, VRK1, VWA1, WARS1, WNK1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend (CMT1, CMT4, HMSN1): 20 Gene (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, axonal (CMT2, HMSN2): 35 Gene (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX, HMSN): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophe Déjerine-Sottas-Neuropathie (DSS, HMSN3): 4 Gene (7,1 kb) EGR2, MPZ, PMP22, PRX Kongenitale hypomyelinisierende Neuropathie (CHN): 3 Gene (6,3 kb) EGR2, MPZ, CNTNAP1 Distale motorische Neuropathie (HMND, HMNR): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Sensorische und autonome Neuropathie (HSAN, HSN): 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	4 - 6 Wo	E
<b>Hereditäre sensorische und autonome Neuropathie (HSAN, HSN) *</b> Gen-Panel: ID086.02, 16 Gene (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	3 - 5 Wo	E
<b>Hirnatrophie und demyelinisierende Erkrankungen des Gehirns *</b> 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
<b>Hyperekplexie (HKPX) *</b> Gen-Panel: ID216.00, 9 Gene (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 Wo	E
<b>Hypomyelinisierende Leukodystrophie (HLD) *</b> 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
<b>Neurodegeneration mit Eisenablagerung im Gehirn (NBIA) *</b> Gen-Panel: ID264.00, 11 Gene (20,0 kb) ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45	3 - 5 Wo	E
<b>Neurologische Entwicklungsstörungen (NED), umfassende Diagnostik *</b> 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, FDF1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, 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, PPF1B1, 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, THUMP1, TIAM1, TMEM147, TMEM222, TMX2, TNF, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 Wo	E
<b>Neuronale Ceroid-Lipofuszinose (CLN) *</b> 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

Erkrankung/Diagnostik	Dauer	Material
<b>Neurodegenerative Erkrankungen</b>		
<p><b>Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik *</b> Gen-Panel: ID204.04</p> <p>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</p> <p>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</p> <p>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</p> <p>Orthochromatische Leukodystrophie: 10 Gene (14,6 kb) ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1</p> <p>Metachromatische Leukodystrophie: 3 Gene (4,2 kb) ARSA, PSAP, SUMF1</p> <p>Aicardi-Goutières-Syndrom (AGS): 7 Gene (11,9 kb) ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1</p> <p>CADASIL, CARASIL: 2 Gene (8,4 kb) HTRA1, NOTCH3</p>	4 - 6 Wo	E
<p><b>Parkinson-Krankheit (PARK) *</b> Gen-Panel: ID077.02</p> <p>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</p> <p>Parkinson-Krankheit (PARK), early-onset: 9 Gene (30,3 kb) ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, SYNJ1, VPS13C</p> <p>Parkinson-Krankheit (PARK), late-onset: 12 Gene (27,3 kb) ADH1C, CHCHD2, EIF4G1, GBA, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, SNCA, UCHL1, VPS35</p> <p>Dystonie-Parkinsonismus (DYT): 9 Gene (18,3 kb) ATP1A3, GCH1, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH</p>	3 - 5 Wo	E
<p><b>Paroxysmale Dyskinesie *</b> Gen-Panel: ID286.00, 14 Gene (38,9 kb)</p> <p>ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24</p>	3 - 5 Wo	E
<p><b>Schlafstörung *</b> Gen-Panel: ID371.00, 13 Gene (25,3 kb)</p> <p>ADRB1, BHLHE41, CRY1, CRY2, CSNK1D, GRM1, HCRT, MOG, NPSR1, PER2, PER3, PRNP, TIMELESS</p>	3 - 5 Wo	E
<p><b>Spastische Ataxie (SPAX) *</b> Gen-Panel: ID228.00, 12 Gene (34,8 kb)</p> <p>AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1</p>	3 - 5 Wo	E
<p><b>Spastische Paraplegie (HSP, SPG) *</b> Gen-Panel: ID148.04</p> <p>Spastische Paraplegie (HSP, SPG): 67 Gene (138,1 kb) ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27</p> <p>Spastische Paraplegie (SPG), autosomal-dominant: 20 Gene (43,1 kb) ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27</p> <p>Spastische Paraplegie (SPG), autosomal-rezessiv: 50 Gene (101,2 kb) ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, UCHL1, VPS37A, ZFYVE26</p> <p>Spastische Paraplegie (SPG), X-chromosomal: 3 Gene (6,9 kb) ABCD1, L1CAM, PLP1</p>	4 - 6 Wo	E
<p><b>Spinale Muskelatrophie (SMA) *</b> Gen-Panel: ID152.02</p> <p>Spinale Muskelatrophie (SMA): 39 Gene (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1</p> <p>Proximale spinale Muskelatrophie (SMA): 13 Gene (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB</p> <p>Distale spinale Muskelatrophie (DSMA, HMN): 24 Gene (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1</p>	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Neurodegenerative Erkrankungen</b>		
<b>Spinozerebelläre Ataxie, autosomal-dominant (SCA, ADCA) *</b> Gen-Panel: ID236.03, 27 Gene (91,6 kb) AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPTX1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2	3 - 5 Wo	E
<b>Spinozerebelläre Ataxie, autosomal-rezessiv (SCAR, SCAN) *</b> Gen-Panel: ID213.04, 36 Gene (117,6 kb) ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PNPLA6, POLG, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TWNK, UBA5, VPS13D, VPS41, VWA3B, WDR73, WWOX, XRCC1	4 - 6 Wo	E
<b>Störung der Peroxisomenbiogenese (PBD) *</b> Gen-Panel: ID083.01 Störung der Peroxisomenbiogenese (PBD): 14 Gene (19,9 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Zellweger-Syndrom (PBD, Typ A): 12 Gene (18,3 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Neonatale Adrenoleukodystrophie / Infantiles Refsum-Syndrom (PBD, Typ B): 11 Gene (17,0 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26 Heimler-Syndrom (PBD, Typ C): 2 Gene (6,8 kb) PEX1, PEX6	3 - 5 Wo	E
<b>Nierenerkrankungen</b>		
<b>Alport-Syndrom (ATS) *</b> Gen-Panel: ID099.00, 4 Gene (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 Wo	E
<b>Aminoazidurie *</b> Gen-Panel: ID318.00 Aminoazidurie: 13 Gene (21,1 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2 Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Renotubuläres Fanconi-Syndrom (FRS): 5 Gene (7,7 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC34A1	3 - 5 Wo	E
<b>Atypisches hämolytisch-urämisches Syndrom (AHUS) *</b> 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
<b>Bardet-Biedl-Syndrom (BBS) *</b> Gen-Panel: ID093.02, 21 Gene (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 Wo	E
<b>Bartter-Syndrom (BARTS) *</b> Gen-Panel: ID156.01, 8 Gene (17,7 kb) BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3	3 - 5 Wo	E
<b>Branchiotorenales Syndrom (BOR) *</b> Gen-Panel: ID315.00, 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 Wo	E
<b>Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen *</b> Gen-Panel: ID706.01, 16 Gene (30,1 kb) ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1	3 - 5 Wo	E
<b>Diabetes insipidus *</b> Gen-Panel: ID322.00, 5 Gene (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 Wo	E
<b>Endokrine Hypertonie *</b> Gen-Panel: ID270.03 Endokrine Hypertonie: 33 Gene (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronismus (HALD): 5 Gene (19,1kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronismus Typ II (PHA2): 4 Gene (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Phäochromozytom-Paragangliom-Syndrom (PPGL): 11 Gene (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenales Cushing-Syndrom (PPNAD): 4 Gene (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle-Syndrom (LIDL): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertonie mit niedrigem Plasma-Renin-Spiegel: 20 Gene (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Nierenerkrankungen</b>		
<b>Fraser-Syndrom (FRASRS) *</b> Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
<b>Galloway-Mowat-Syndrom (GAMOS) *</b> Gen-Panel: ID251.01, 10 Gene (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 Wo	E
<b>Glomerulonephritis *</b> Gen-Panel: ID103.01 Glomerulonephritis: 18 Gene (36,9 kb) C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD C3-Glomerulopathie (C3G): 3 Gene (7,2 kb) CFI, CFH, CFHR5 C1q-Mangel (C1QD): 3 Gene (2,3 kb) C1QA, C1QB, C1QC	3 - 5 Wo	E
<b>Hyperoxalurie</b> Gen-Panel: ID363.00 Hyperoxalurie: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb) AGXT, GRHPR, HOGA1 Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 Wo	E
<b>Hypomagnesiämie (HOMG) *</b> Gen-Panel ID054.02: 14 Gene (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXRD2, HNF1B, KCNA1, KCNJ10, RRRAGD, SLC12A3, TRPM6	3 - 5 Wo	E
<b>Hypophosphatasie, Hypophosphatämie und Rachitis *</b> 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
<b>Joubert-Syndrom (JBTS) *</b> Gen-Panel: ID028.03, 40 Gene (104,1 kb) AH11, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 Wo	E
<b>Kongenitale Anomalien der Niere und ableitenden Harnwege (CAKUT) *</b> Gen-Panel: ID229.03 Kongenitale Anomalien der Niere und ableitenden Harnwege (CAKUT): 62 Gene (198,3 kb) ACE, ACTG2, AGT, AGTR1, ANOS1, BICC1, BMP4, BNC2, CDC5L, CEP55, CHD1L, CHRM3, CRKL, DSTYK, EYA1, FAT4, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3, GFRA1, GREB1L, GRIP1, HNF1B, HPSE2, ITGA8, KIF14, LIFR, LMOD1, LRIG2, LRP4, MUC1, MYH11, MYL9, MYLK, NEK8, NPHP3, NRIP1, PAX2, PBX1, REN, RET, ROBO1, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX11, SOX17, TBC1D1, TBX18, TFAP2A, TNXB, TRAP1, UMOD, UPK3A, WBP11, WNT4 Renale Hypodysplasie/Aplasie und Agenesie: 25 Gene (75,9 kb) ANOS1, BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GFRA1, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4 Vesikoureteraler Reflux (VUR): 10 Gene (33,7 kb) DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB Branchiotorenales Syndrom (BOR): 5 Gene (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A Renale tubuläre Dysgenese (RTD): 4 Gene (7,7 kb) ACE, AGT, AGTR1, REN Fraser-Syndrom (FRASRS): 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
<b>Meckel-Syndrom (MKS) *</b> 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
<b>Metabolische Nierenerkrankungen *</b> 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
<b>Mikrohämaturie *</b> Gen-Panel: ID385.00, 11 Gene (44,0 kb) CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FN1, INF2, MYH9, PIGA, PIGT, UMOD	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Nierenerkrankungen</b>		
<b>Nephrokalzinose *</b> 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, GRHRP, HOGA1, KCNJ1, MAGED2, OCRL, OXGR1, RRAGD, SLC12A1, SLC4A1, SLC34A1, SLC34A3, VIPAS39, VPS33B	3 - 5 Wo	E
<b>Nephronophthie (NPHP) *</b> Gen-Panel: ID030.02, 22 Gene (70,9 kb) ANKS6, CEP83, CEP164, CEP290, DDCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	3 - 5 Wo	E
<b>Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS) *</b> Gen-Panel: ID098.06 Nephrotisches Syndrom (SRNS, NPHS) und Fokal-segmentale Glomerulosklerose (FSGS): 43 Gene (129,7 kb) ACTN4, ANLN, APOL1, ARHGAP24, ARHGADIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, DAAM2, DGKE, EMP2, INF2, KANK2, KIRREL1, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PAX2, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, TRPC6, WT1 Nephrotisches Syndrom (SRNS, NPHS): 30 Gene (89,3 kb) ARHGAP24, ARHGADIA, AVIL, COQ2, COQ6, COQ8B, DAAM2, DGKE, EMP2, KANK2, KIRREL1, LAMA5, LAMB2, MAGI2, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, WT1 Fokal-segmentale Glomerulosklerose (FSGS): 18 Gene (49,5 kb) ACTN4, ANLN, APOL1, ARHGAP24, COQ2, COQ6, COL4A3, COL4A4, COL4A5, CRB2, CD2AP, INF2, LMX1B, MYO1E, PAX2, PDSS2, SMARCAL1, TRPC6	4 - 6 Wo	E
<b>Nierenzellkarzinom *</b> 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
<b>Polyzystische Lebererkrankung (PCLD) *</b> Gen-Panel: ID305.02 Polyzystische Lebererkrankung (PCLD): 16 Gene (55,7 kb) ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63 Polyzystische Lebererkrankung mit oder ohne Nierenzysten (PCLD): 7 Gene (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Polyzystische Nierenerkrankung mit polyzystischer Lebererkrankung (PKD): 9 Gene (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1	3 - 5 Wo	E
<b>Polyzystische Nierenerkrankung (PKD) *</b> Gen-Panel: ID295.03 Polyzystische Nierenerkrankung (PKD): 9 Gene (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1 Polyzystische Nierenerkrankung, autosomal-dominant (ADPKD): 7 Gene (27,4 kb) ALG5, DNAJB11, GANAB, IFT140, NEK8, PKD1, PKD2 Polyzystische Nierenerkrankung, autosomal-rezessiv (ARPKD): 2 Gene (14,5 kb) DZIP1L, PKHD1	3 - 5 Wo	E
<b>Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA) *</b> Gen-Panel: ID250.01 Pseudoaldosteronismus (LIDLS) und Pseudohypoaldosteronismus (PHA): 8 Gene (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronismus, Typ I (PHA1): 4 Gene (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronismus, Typ II (PHA2): 4 Gene (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle-Syndrom (LIDLS): 3 Gene (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 Wo	E
<b>Renale Amyloidose *</b> Gen-Panel ID320.01 Renale Amyloidose: 9 Gene (13,2 kb) APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 6 Gene (6,3 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Periodisches Fiebersyndrom mit Amyloidose: 3 Gene (6,8 kb) MEFV, NLRP3, TNFRSF1A	3 - 5 Wo	E
<b>Renale Hypodysplasie, Aplasie und Agenesie *</b> Gen-Panel: ID319.00, 23 Gene (72,5 kb) BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4	3 - 5 Wo	E
<b>Renale tubuläre Azidose (RTA) *</b> Gen-Panel: ID297.00, 9 Gene (18,5 kb) ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72	3 - 5 Wo	E
<b>Renale tubuläre Dysgenese (RTD) *</b> Gen-Panel: ID316.00, 4 Gene (7,7 kb) ACE, AGT, AGTR1, REN	2 - 4 Wo	E
<b>Renotubuläres Fanconi-Syndrom (FRTS) *</b> Gen-Panel: ID359.00, 7 Gene (10,4 kb) CTNS, EHHADH, GATM, HNF4A, NDUFAF6, SLC2A2, SLC34A1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Nierenerkrankungen</b>		
<b>Renale Zilopathien, umfassende Diagnostik *</b> Gen-Panel: ID376.00 Renale Zilopathien, umfassende Diagnostik: 75 Gene (181,7 kb) AHI1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPGRIP1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, XPNPEP3, ZNF423 Nephronophthuse (NPHP): 17 Gene (50,3 kb) ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP Senior-Loken-Syndrom (SLSN): 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 Joubert-Syndrom (JBTS): 40 Gene (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423 Meckel-Syndrom (MKS): 14 Gene (36,2 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15	4 - 6 Wo	E
<b>Senior-Loken-Syndrom (SLSN) *</b> Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E
<b>Tubulointerstitielle Nierenerkrankung, autosomal-dominant (ADTKD) *</b> Gen-Panel: ID296.00, 6 Gene (8,8 kb) DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD	3 - 5 Wo	E
<b>Urolithiasis, Nephrolithiasis und Nephrokalzinose *</b> Gen-Panel ID231.04 Urolithiasis, Nephrolithiasis und Nephrokalzinose: 37 Gene (68,8 kb) ADCY10, AGXT, ALPL, APRT, ATP6VOA4, ATP6V1B1, BSND, CASR, CLCN5, CLDN16, CLDN19, CLCNKB, CYP24A1, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, OCRL, OXGR1, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, XDH Hyperkalziurie: 15 Gene (30,2 kb) ADCY10, BSND, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, RRAGD, SLC12A1, SLC34A1, SLC34A3 Hyperoxalurie: 5 Gene (6,3 kb) AGXT, GRHPR, HOGA1, SLC26A1, OXGR1 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Hypocitraturie: 4 Gene (11,1 kb) TP6VOA4, ATP6V1B1, SLC4A1, SLC4A4 Xanthinurie: 2 Gene (6,7 kb) MOCOS, XDH Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9	3 - 5 Wo	E
<b>Urothelkarzinom *</b> Gen-Panel: ID337.00, 34 Gene (105,0 kb) APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC	3 - 5 Wo	E
<b>Vesikoureteraler Reflux (VUR) *</b> Gen-Panel: ID314.00, 10 Gene (33,7 kb) DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB	3 - 5 Wo	E
<b>Wilms-Tumor (WT) *</b> 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	3 - 5 Wo	E
<b>Zystische Nierenerkrankungen, umfassende Diagnostik *</b> Gen-Panel: ID100.09 Zystische Nierenerkrankungen, umfassende Diagnostik: 53 Gene (165,3 kb) ALG5, ALG6, ALG8, ALG9, ANKS6, CEP83, CEP164, CEP290, COL4A1, CRB2, CYS1, DCDC2, DNAJB11, DZIP1L, FLCN, GANAB, GLIS2, HNF1B, IFT140, IFT172, INVS, IQCB1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, REN, RPGRIP1L, SDCCAG8, SEC61A1, SEC61B, SEC63, SLC41A1, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, XPNPEP3, ZNF423 Polyzystische Nierenerkrankung (PKD): 9 Gene (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1 Polyzystische Lebererkrankung mit Nierenzysten (PCLD): 7 Gene (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Medulläre zystische Nierenerkrankung (MCKD, ADTKD): 5 Gene (7,7 kb) HNF1B, MUC1, REN, SEC61A1, UMOD Nephronophthuse (NPHP): 17 Gene (50,3 kb) ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Skelett- und Knochenkrankungen</b>		
<b>Adams-Oliver-Syndrom (AOS) *</b> Gen-Panel: ID259.00, 6 Gene (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 Wo	E
<b>Akrozephalosyndaktylie (ACS) *</b> Gen-Panel: ID311.00, 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1	3 - 5 Wo	E
<b>Arachnodaktylie *</b> Gen-Panel: ID124.00, 13 Gene (35,9 kb) CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFB3, TGFB3, SMAD3	3 - 5 Wo	E
<b>Arthrogrypose *</b> Gen-Panel: ID200.01 Arthrogrypose: 61 Gene (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNM2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSIN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2 Arthrogryposis multiplex congenita (AMC): 6 Gene (58,2 kb) ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1 Distale Arthrogrypose (DA): 11 Gene (40,3 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1 Letales kongenitales Kontraktursyndrom (LCCS): 11 Gene (31,5 kb) ADCY6, ADGRG6, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42 Fetale Akinesie-Deformation-Sequenz (FADS): 4 Gene (7,6 kb) DOK7, MUSK, NUP88, RAPSIN	4 - 6 Wo	E
<b>Brachydaktylie (BD) *</b> 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
<b>Distale Arthrogrypose (DA) *</b> Gen-Panel: ID196.02, 11 Gene (40,5 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1	3 - 5 Wo	E
<b>Fraser-Syndrom (FRASRS) *</b> Gen-Panel: ID317.00, 3 Gene (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 Wo	E
<b>Frontonasale Dysplasie (FND) *</b> Gen-Panel ID339.00: 11 Gene (22,1 kb) ALX1, ALX3, ALX4, ANKH, EFN1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6	3 - 5 Wo	E
<b>Handfehlbildungen, umfassende Diagnostik *</b> Gen-Panel: ID298.00 Handfehlbildungen, umfassende Diagnostik: 110 Gene (295,9 kb) ADAMTS10, ADAMTS17, AKT3, BHLHA9, BMP2, BMPR1B, C2CD3, CACNA1C, CCND2, CCNQ, CDH3, CHST11, CHSY1, CIBAR1, CKAP2L, CPLANE1, CREBBP, DACT1, DDX59, DHCR7, DHODH, DLL4, DLX5, DOCK6, EFN1, EFTUD2, EOGT, EP300, ESCO2, EVC2, FBLN1, FBN1, FGF10, FGF16, FGF9, 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, SMOG1, SOST, TBC1D24, TBX15, TBX3, TBX5, TCTN3, TMEM107, TP63, TRPV4, TWIST1, WDCPCP, WNT10B, WNT7A, YY1AP1, ZNF141 Brachydaktylie (BD), nicht-syndromal: 9 Gene (14,0 kb) BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PDE3A, PTHLH, ROR2 Polydaktylie (PAPA, PPD), nicht-syndromal: 9 Gene (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141 Syndaktylie (SDTY), nicht-syndromal: 8 Gene (18,5 kb) BHLHA9, FBLN1, GJA1, GLI3, HOXD13, LMBR1, LRP4, NECTIN4 Ektrodaktylie (SHFM): 7 Gene (10,6 kb) CDH3, DLX5, FGFR1, IGF2, TP63, WNT7A, WNT10B Akrozephalosyndaktylie (ACS): 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Orofaziodigitales Syndrom (OFD): 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107 Lakrimoaurikulodentodigitales Syndrom (LADD): 3 Gene (5,5 kb) FGF10, FGFR2, FGFR3 Multiple Synostosen-Syndrom (SYNS): 6 Gene (8,3 kb) FGF9, GDF5, GDF6, HOXA11, MECOM, NOG Akrodysostosis-Syndrom (ACRDYS): 3 Gene (4,9 kb) PDE4D, PRKAR1A, SF3B4	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Skelett- und Knochenkrankungen</b>		
<b>Herz-Hand-Syndrom *</b> Gen-Panel: ID165.01, 9 Gene (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 Wo	E
<b>Hypophosphatasie, Hypophosphatämie und Rachitis *</b> 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
<b>Klippel-Feil-Syndrom (KFS) *</b> Gen-Panel: ID207.00, 5 Gene (12,5 kb) GDF3, GDF6, MEOX1, MYO18B, PAX1	3 - 5 Wo	E
<b>Kraniosynostose (CRS) *</b> Gen-Panel: ID224.02 Kraniosynostose (CRS): 36 Gene (84,9 kb) ALPL, ALX4, ASXL1, CD96, CDC45, COLEC10, COLEC11, CYP26B1, EFN1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT43, IFT122, IL11RA, MASP1, MEGF8, MSX2, P4HB, POR, PPP3CA, RAB23, RECQL4, SCARF2, SEC24D, SKI, SMAD6, TCF12, TWIST1, WDR19, WDR35, ZIC1 Kraniosynostose (CRS), nicht-syndromal: 8 Gene (10,5 kb) ALX4, ERF*, IL11RA, MSX2, SMAD6, TCF12, TWIST1, ZIC1 Akrozephalosyndaktylie (ACS): 6 Gene (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Kranioektodermale Dysplasie (CED): 4 Gene (12,1 kb) IFT43, IFT122, WDR19, WDR35 Trigonozephalie (TRIGNO): 5 Gene (17,0 kb) ASXL1, CD96, FGFR1, FREM1, PPP3CA	3 - 5 Wo	E
<b>Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD) *</b> Gen-Panel: ID067.00 Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD): 20 Gene (68,2 kb) CEP120, DYNC2H1, DYNC2L1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Kurzrippen-Thoraxdysplasie (Jeune-ATD, SRPS, SRTD): 18 Gene (61,3 kb) CEP120, DYNC2H1, DYNC2L1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Ellis-van-Crefeld-Syndrom (EVC): 4 Gene (11,5 kb) DYNC2L1, EVC, EVC1, WDR35	3 - 5 Wo	E
<b>Letales kongenitales Kontraktursyndrom (LCCS) *</b> 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
<b>Lippen-, Kiefer- und Gaumenspalte (OFC, CLP) *</b> Gen-Panel: ID266.01 Lippen-, Kiefer- und Gaumenspalte (OFC, CLP): 50 Gene (151,0 kb) ARHGAP29, BMP4, BRD4, C2CD3, CDH1, CHD7, CDH11, CPLANE1, CYFIP1, DDX59, DHCR7, DLX4, ESCO2, FGFR1, FGFR2, FLNB, GRHL3, HDAC8, IFT57, INTU, IRF6, KDM1A, KDM6A, KIAA0753, KMT2D, LRRC32, MEIS2, MID1, MSX1, NECTIN1, NEK1, NIPA1, NIPA2, NIPBL, OFD1, RAD21, RAB34, RIPK4, SCNM1, SLC26A2, SMC1A, SMC3, SPECC1L, TBX22, TCTN3, TGDS, TMEM107, TP63, TUBGCP5, ZRSR2 Orofaziale Spalte, nicht-syndromal (OFC): 9 Gene (15,1 kb) ARHGAP29, BMP4, DLX4, GRHL3, IRF6, MSX1, NECTIN1, TBX22, TP63 Orofaziodigitales Syndrom (OFD): 13 Gene (36,5 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCNM1, TCTN3, TMEM107, ZRSR2 Cornelia-de-Lange-Syndrom (CDLS): 6 Gene (22,9 kb) BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3 Kabuki-Syndrom (KABUK): 3 Gene (23,5 kb) KDM1A, KDM6A, KMT2D Van-der-Woude-Syndrom (VWS): 2 Gene (3,3 kb) GRHL3, IRF6	4 - 6 Wo	E
<b>Mandibulofaziale Dysostose (MFD) *</b> 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
<b>Multiple epiphysäre Dysplasie (EDM) *</b> Gen-Panel: ID202.02, 11 Gene (27,7 kb) CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CSGALNACT1, EIF2AK3, KIF7, MATN3, SLC26A2	3 - 5 Wo	E
<b>Multiples Pterygium-Syndrom *</b> Gen-Panel: ID158.01, 8 Gene (16,8 kb) CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	3 - 5 Wo	E
<b>Orofaziodigitales Syndrom (OFD) *</b> Gen-Panel: ID265.00, 9 Gene (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Skelett- und Knochenkrankungen</b>		
<b>Osteogenesis imperfecta (OI) *</b> 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
<b>Osteopetrose (OPT) und ähnliche Knochenkrankungen *</b> Gen-Panel ID346.01 Osteopetrose (OPT) und ähnliche Knochenkrankungen: 32 Gene (68,1 kb) AMER1, ANKH, CA2, CLCN7, CSF1R, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, LRP6, LRRK1, OSTM1, PLEKHM1, PTSS1, PTH1R, SLC4A2, SLC29A3, SLC02A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP Osteopetrose (OPTA, OPTB): 13 Gene (30,2 kb) CA2, CLCN7, FERMT3, LRP5, LRP6, OSTM1, PLEKHM1, SLC4A2, SLC29A3, SNX10, TCIRG1, TNFRSF11A, TNFSF11 Syndromale Skelettdysplasie mit erhöhter Knochendichte: 19 Gene (38,0 kb) AMER1, ANKH, CSF1R, CTSK, DLX3, FAM20C, GJA1, HPGD, LEMD3, LRP4, LRRK1, PTSS1, PTH1R, SLC02A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP	3 - 5 Wo	E
<b>Osteoporose *</b> Gen-Panel: ID115.01, 13 Gene (29,4 kb) CALCR, COL1A1, COL1A2, ESR1, LRG4, LRP5, PLS3, SGMS2, SLC34A1, SLC9A3R1, UGT2B17, VDR, WNT1	3 - 5 Wo	E
<b>Pierre-Robin-Sequenz *</b> Gen-Panel: ID294.00, 33 Gene (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 Wo	E
<b>Polydaktylie, nicht-syndromale Form *</b> Gen-Panel: ID166.02, 9 Gene (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141	3 - 5 Wo	E
<b>Rubinstein-Taybi-Syndrom (RSTS) *</b> Gen-Panel: ID142.01, 3 Gene (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 Wo	E
<b>Skelettdysplasie, schwere Form *</b> 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 Achondrogenese (ACG): 4 Gene (14,1 kb) COL2A1, GDF5, SLC26A2, TRIP11 Fibrochondrogenese (FBCG): 2 Gene (10,6 kb) COL11A1, COL11A2 Thanatophore Dysplasie (TD): 2 Gene (6,9 kb) COL1A2, FGFR3 Chondrodysplasie, letal: 12 Gene (24,5 kb) AGPS, EBP, FLNB, GDF5, GNPAT, GPX4, LBR, PEX5, PEX7, PTH1R, SLC26A2, SLC35D1 Osteogenesis imperfecta (OI), letal: 5 Gene (12,6 kb) CRTAP, COL1A1, COL1A2, P3H1, PPIB Kurzrippen-Thoraxdysplasie (SRTD), letal: 9 Gene (35,0 kb) CEP120, DYNC2H1, IFT52, IFT80, IFT81, KIAA0586, NEK1, WDR34, WDR35	4 - 6 Wo	E
<b>Skelettdysplasien, umfassende Diagnostik *</b> Gen-Panel: ID356.00, 407 Gene (969,6 kb) ABCG9, 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, CCN9, 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, DYNC2L1, 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, PTSS1, 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, SLC02A1, SMAD3, SMAD4, SMAD6, SMARCA1, 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, TGFB3, TMC01, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UFPS2, UNC45A, VDR, WBP11, WPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6	4 - 6 Wo	E
<b>Spondylkostale Dysostose (SCDO) *</b> Gen-Panel: ID227.00, 7 Gene (14,4 kb) DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Skelett- und Knochenkrankungen</b>		
<b>Spondyloepiphysäre und spondylometaphysäre Dysplasie (SED, SMD, SEMD) *</b> 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
<b>Weill-Marchesani-Syndrom (WMS) *</b> Gen-Panel: ID230.00, 4 Gene (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wo	E
<b>3M-Syndrom *</b> Gen-Panel: ID214.00, 3 Gene (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 Wo	E
<b>Stoffwechselerkrankungen</b>		
<b>Aminoazidurie *</b> Gen-Panel: ID318.00 Aminoazidurie: 13 Gene (21,1 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2 Cystinurie: 2 Gene (3,5 kb) SLC3A1, SLC7A9 Hyperglycinurie: 3 Gene (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Renotubuläres Fanconi-Syndrom (FRTS): 5 Gene (7,7 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC34A1	3 - 5 Wo	E
<b>Amyloidose *</b> Gen-Panel: ID375.00 Amyloidose: 18 Gene (25,0 kb) APP, APOA1, APOA2, APOC2, B2M, CST3, FGA, GPNMB, GSN, IL31RA, ITM2B, LYZ, MEFV, NLRP3, OSMR, PRNP, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 8 Gene (7,0 kb) APOA1, APOA2, APOC2, B2M, FGA, GSN, LYZ, TTR Primäre lokalisierte kutane Amyloidose (PLCA): 3 Gene (7,0 kb) GPNMB, OSMR, IL31RA Zerebrale Amyloidangiopathie (HCHWA): 4 Gene (4,3 kb) APP, CST3, ITM2B, PRNP	3 - 5 Wo	E
<b>Coenzym-Q10-Mangel (COQ10D) *</b> 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
<b>Cystinose (CTNS) und ähnliche Stoffwechselerkrankungen *</b> Gen-Panel: ID706.01, 16 Gene (30,1 kb) ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1	3 - 5 Wo	E
<b>Folatstoffwechselstörung *</b> Gen-Panel: ID334.00, 10 Gene (18,7 kb) CBS*, FOLR1, FOLR2, FTCD, MTHFD1, MTHFR, MTR, MTRR, SLC19A1, SLC46A1	3 - 5 Wo	E
<b>Glykogenspeicherkrankheit (GSD) *</b> Gen-Panel: ID108.01, 29 Gene (54,1 kb) AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC1, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4	3 - 5 Wo	E
<b>Glykosylphosphatidylinositol (GPI)-Biosynthesedefekt (GPIBD) *</b> Gen-Panel: ID291.00 Glykosylphosphatidylinositol(GPI)-Biosynthesedefekt (GPIBD): 22 Gene (33,3 kb) GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY Multiple kongenitale Anomalien-Hypotonie-Krampfanfälle-Syndrom (MCAHS): 4 Gene (7,7 kb) PIGA, PIGN, PIGQ, PIGT Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS): 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
<b>Hämochromatose (HFE) und Häm siderose *</b> Gen-Panel: ID114.04, 10 Gene (14,6 kb) BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2	3 - 5 Wo	E
<b>Homocystinurie *</b> Gen-Panel: ID191.01, 9 Gene (15,3 kb) ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Stoffwechselerkrankungen</b>		
<b>Hyperinsulinämische Hypoglykämie (HHF) *</b> Gen-Panel: ID126.00, 8 Gene (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 Wo	E
<b>Hyperkalzämie *</b> Gen-Panel: ID262.00, 8 Gene (14,6 kb) AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1	3 - 5 Wo	E
<b>Hyperoxalurie</b> Gen-Panel: ID363.00 Hyperoxalurie: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primäre Hyperoxalurie (PH): 3 Gene (3,2 kb) AGXT, GRHPR, HOGA1 Kalziumoxalat-Nephrolithiasis (CAON): 3 Gene (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 Wo	E
<b>Hyperphosphatasie-Intelligenzminderung-Syndrom (HPMRS) *</b> Gen-Panel: ID292.00, 6 Gene (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 Wo	E
<b>Hypoglykämie, Hyperinsulinismus und Ketonstoffwechselstörung *</b> 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
<b>Hypomagnesiämie (HOMG) *</b> Gen-Panel ID054.02: 14 Gene (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6	3 - 5 Wo	E
<b>Hypophosphatasie, Hypophosphatämie und Rachitis *</b> 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
<b>Kombinierter Defekt der oxidativen Phosphorylierung (COXPD) *</b> Gen-Panel: ID287.01, 60 Gene (80,3 kb) AARS2, AIFM1, ATP5F1A, C10BP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 Wo	E
<b>Kongenitale Störung der Glykosylierung (CDG) *</b> 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, ATP6VOA2, 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, ATP6VOA2, 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
<b>Muskeldystrophie-Dystroglukanopathie (MDDG) *</b> Gen-Panel: ID179.00 Muskeldystrophie-Dystroglukanopathie (MDDG): 15 Gene (24,0 kb) B3GALNT2, B4GAT1, DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muskeldystrophie-Dystroglukanopathie (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-Dystroglukanopathie (MDDGB) mit oder ohne intellektuelle Entwicklungsstörung: 8 Gene (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2 Muskeldystrophie-Dystroglukanopathie (MDDGC), Gliedergürtelmuskeldystrophie: 11 Gene (18,5 kb) DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, POMGNT1, POMGNT2, POMK, POMT1, POMT2	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Stoffwechselerkrankungen</b>		
<b>Metabolische Epilepsien *</b> Gen-Panel: ID303.01 Metabolische Epilepsien: 84 Gene (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, CLN3, CLN5, CLN6, CLN8, CNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETF, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycin-Enzephalopathie (GCE): 5 Gene (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdän-Cofaktor-Defizienz (MOCOD): 3 Gene (3,7 kb) GPHN, MOCS1, MOCS2 Zerebrales Kreatinmangelsyndrom (CCDS): 3 Gene (3,9 kb) GAMT, GATM, SLC6A8 Ahornsirupkrankheit (MSUD): 5 Gene (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxy-Glutarazidurie: 3 Gene (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-Gangliosidose: 3 Gene (3,8 kb) HEXA, HEXB, GM2A Neuronale Ceroid-Lipofuszinose (CLN): 12 Gene (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 Wo	E
<b>MODY-Diabetes *</b> 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
<b>Mukopolysaccharidose (MPS) *</b> Gen-Panel: ID308.00, 12 Gene (21,2 kb) ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A	3 - 5 Wo	E
<b>Neonataler Diabetes mellitus *</b> 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
<b>Neuronale Ceroid-Lipofuszinose (CLN) *</b> 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
<b>Porphyrie *</b> Gen-Panel: ID153.01, 10 Gene (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 Wo	E
<b>Pulmonale Surfactant-Stoffwechselstörung (SMDP) *</b> Gen-Panel: ID168.01, 6 Gene (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 Wo	E
<b>Renale Amyloidose *</b> Gen-Panel ID320.01 Renale Amyloidose: 9 Gene (13,2 kb) APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TNFRSF1A, TTR Hereditäre systemische Amyloidose (AMYLD): 6 Gene (6,3 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Periodisches Fiebersyndrom mit Amyloidose: 3 Gene (6,8 kb) MEFV, NLRP3, TNFRSF1A	3 - 5 Wo	E
<b>Speicherkrankheiten mit Herzbeteiligung *</b> Gen-Panel: ID149.02 Speicherkrankheiten mit Herzbeteiligung: 17 Gene (25,1 kb) APOA1, ATP7B, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Amyloidose (AMYLD): 6 Gene (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Hämochromatose (HFE): 6 Gene (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Kardiale Glykogenspeicherkrankheit (GSD): 3 Gene (5,8 kb) GAA, LAMP2, PRKAG2	3 - 5 Wo	E
<b>Stoffwechselstörung mit Epilepsie im Neugeborenenalter *</b> 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
<b>Stoffwechselstörung mit Epilepsie im Säuglings-, Kleinkind- und Schulalter *</b> 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



Erkrankung/Diagnostik	Dauer	Material
<b>Tumorerkrankungen</b>		
<b>Endometriumkarzinom, umfassende Diagnostik *</b> Gen-Panel: ID365.00, 26 Gene (92,9 kb) ATM, APC, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11, TP53	3 - 5 Wo	E
<b>Fanconi-Anämie (FANC) *</b> Gen-Panel: ID043.02, 21 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFW3, SLX4, UBE2T, XRCC2	3 - 5 Wo	E
<b>Gastrointestinaler Stromatumor (GIST) *</b> Gen-Panel: ID226.00, 8 Gene (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 Wo	E
<b>Glioblastom (GLM) *</b> 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
<b>Gorlin-Goltz-Syndrom (BCNS) und ähnliche Krankheitsbilder *</b> Gen-Panel: ID174.01, 7 Gene (23,6 kb) CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 Wo	E
<b>Hypophysenadenom (PITA) *</b> Gen-Panel: ID387.00, 8 Gene (20,7 kb) AIP, CDH23, CDKN1B, GNAS, GPR101, MEN1, PRKAR1A, USP8	3 - 5 Wo	E
<b>Kolorektales Karzinom *</b> Gen-Panel: ID049.03, 18 Gene (55,7 kb) APC, ATM, BMPR1A, CHEK2, EPCAM, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11	3 - 5 Wo	E
<b>Kolorektales Karzinom, umfassende Diagnostik *</b> Gen-Panel: ID285.02, 34 Gene (89,7 kb) APC, ATM, AXIN2, BLM, BMPR1A, CDH1, CHEK2, EPCAM, EXO1, FLCN, GALNT12, GREM1, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PLA2G2A, PMS2, POLD1, POLE, PTEN, RFC1, RPA1, RNF43, RPS20, SMAD4, STK11, TGFB2, TP53	3 - 5 Wo	E
<b>Kolorektales Karzinom und Polyposis *</b> Gen-Panel ID006.09 Kolorektales Karzinom und Polyposis: 23 Gene (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Kolorektales Karzinom: 18 Gene (55,7 kb) APC, ATM, CHEK2, BMPR1A, EPCAM, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11 Kolorektale Polyposis: 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
<b>Kolorektales Karzinom mit Mikrosatelliteninstabilität (MSI) *</b> Gen-Panel: ID283.00, 9 Gene (25,4 kb) EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE	3 - 5 Wo	E
<b>Konstitutionelles MMR-Defizienz-Syndrom (CMMRDS, MMRCS) *</b> Gen-Panel: ID362.00, 5 Gene (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	2 - 4 Wo	E
<b>Krebserkrankungen im Kindesalter *</b> 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, RFW3, RIT1, RRS2, 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, RFW3, 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, RRS2, 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
<b>Kutanes malignes Melanom (CMM) *</b> 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
<b>Tumorerkrankungen</b>		
<b>Lungenkarzinom *</b> 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
<b>Lynch-Syndrom (LYNCH, HNPCC) *</b> Gen-Panel: ID002.02, 5 Gene (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	2 - 4 Wo	E
<b>Magenkarzinom *</b> Gen-Panel: ID090.04, 25 Gene (73,6 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	3 - 5 Wo	E
<b>Mammakarzinom *</b> 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
<b>Mamma- und Ovarialkarzinom (HBOC) *</b> Gen-Panel: ID003.04 Mamma- und Ovarialkarzinom (HBOC): 19 Gene (62,5 kb) ATM, BARD1, BRIP1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53 Mammakarzinom: 10 Gene (38,9 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, STK11, TP53 Ovarialkarzinom: 14 Gene (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 Wo	E
<b>Mamma- und Ovarialkarzinom, umfassende Diagnostik *</b> 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	4 - 6 Wo	E
<b>Medulloblastom (MDB) *</b> Gen-Panel: ID205.02, 22 Gene (75,7 kb) APC, BRCA2, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTCH1, PTCH2, PTEN, SMARCB1, SMARCA4, SUFU, TP53, VHL	3 - 5 Wo	E
<b>Myelodysplastisches Syndrom (MDS) und Akute myeloische Leukämie (AML) *</b> 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, GF11, 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, RFW3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRS2, 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, GF11, HAX1, JAGN1, SRP54, VPS45, WAS Fanconi-Anämie (FAN): 20 Gene (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFW3, SLX4, UBE2T, XRCC2 Mismatch-Reparatur-Defizienz (CMMRDS, MMRCS): 4 Gene (11,8 kb) MLH1, MSH2, MSH6, PMS2	4 - 6 Wo	E
<b>Neuroendokrine Neoplasie *</b> Gen-Panel: ID386.00, 19 Gene (29,2 kb) AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL	3 - 5 Wo	E
<b>Neurofibromatose (NF) *, #</b> Gen-Panel: ID210.00, 3 Gene (11,6 kb) NF1, NF2, SPRED1	3 - 5 Wo	E
<b>Nierenzellkarzinom *</b> 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
<b>Ovarialkarzinom *</b> Gen-Panel: ID004.04, 14 Gene (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Tumorerkrankungen</b>		
<b>Pankreaskarzinom *</b> 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
<b>Phäochromozytom-Paragangliom-Syndrom (PPGL) *</b> Gen-Panel: ID042.03, 14 Gene (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 Wo	E
<b>Plasmozytom *</b> 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, KLHC8B, KRAS, LAPTM5, MLH1, MSH2, MSH6, MYD88, NBN, NF1, NRAS, PAX5, PRF1, PMS2, POT1, PRKCD, PTPN11, RBM8A, SBDS, SH2B3, SH2D1A, TP53, USP45, WAS	3 - 5 Wo	E
<b>Polyposis-Syndrom (PS, FAP) *</b> Gen-Panel: ID005.06, 15 Gene (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 Wo	E
<b>Prostatakarzinom *</b> Gen-Panel: ID140.03, 30 Gene (101,5 kb) AR, ATM, ATR, BAP1, BARD1, BRCA1, BRCA2, BRIP1, CYP3A43, CDH1, CHEK2, ELAC2, EPCAM, FANCM, HOXB13, MLH1, MRE11, MSR1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, RNASEL, TP53, TRRAP	3 - 5 Wo	E
<b>Sarkom der Weichteile und des Skelettsystems *</b> Gen-Panel: ID223.02, 55 Gene (155,1 kb) APC, ATM, ATR, ATRX, BLM, BRCA2, BUB1B, CDKN1C, CDKN2A, CEP57, CHEK2, DICER1, DKC1, EPCAM, ERCC2, EXT1, EXT2, FAH, FANCC, FH, HRAS, KIT, MEN1, MLH1, MRE11, MSH2, MSH6, MTAP, NBN, NF1, PALB2, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RPS19, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SQSTM1, SUFU, TBXT, TNFRSF11A, TP53, VHL, WAS, WRN, ZNF687	4 - 6 Wo	E
<b>Schilddrüsenkarzinom *</b> Gen-Panel: ID220.02, 26 Gene (60,5 kb) ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TIN2, TP53	3 - 5 Wo	E
<b>Tuberöse Sklerose (TSC) *</b> Gen-Panel: ID332.00, 2 Gene (8,9 kb) TSC1, TSC2	2 - 4 Wo	E
<b>Urothelkarzinom *</b> Gen-Panel: ID337.00, 34 Gene (105,0 kb) APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC	3 - 5 Wo	E
<b>Wilms-Tumor (WT) *</b> 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	3 - 5 Wo	E
<b>Xeroderma pigmentosum (XP) *</b> Gen-Panel: ID282.00, 10 Gene (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 Wo	E
<b>Ziliopathien</b>		
<b>Bardet-Biedl-Syndrom (BBS) *</b> Gen-Panel: ID093.02, 21 Gene (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 Wo	E
<b>Joubert-Syndrom (JBTS) *</b> Gen-Panel: ID028.03, 40 Gene (104,1 kb) AH11, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 Wo	E
<b>Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD) *</b> Gen-Panel: ID067.00 Kurzrippen-Thoraxdysplasie mit oder ohne Polydaktylie (SRTD): 20 Gene (68,2 kb) CEP120, DYNC2H1, DYNC2L1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Kurzrippen-Thoraxdysplasie (Jeune-ATD, SRPS, SRTD): 18 Gene (61,3 kb) CEP120, DYNC2H1, DYNC2L1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Ellis-van-Crefeld-Syndrom (EVC): 4 Gene (11,5 kb) DYNC2L1, EVC, EVC1, WDR35	3 - 5 Wo	E
<b>Meckel-Syndrom (MKS) *</b> 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
<b>Nephronophthie (NPHP) *</b> 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
<b>Senior-Loken-Syndrom (SLSN) *</b> Gen-Panel: ID029.01, 8 Gene (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wo	E

Erkrankung/Diagnostik	Dauer	Material
<b>Ziliopathien</b>		
<b>Primäre Ziliendyskinesie mit oder ohne Situs inversus (PCD, CILD) *</b> Gen-Panel: ID085.03, 50 Gene (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 Wo	E
<b>Renale Ziliopathien, umfassende Diagnostik *</b> Gen-Panel: ID376.00 <b>Renale Ziliopathien, umfassende Diagnostik: 75 Gene (181,7 kb)</b> AH11, ANKS6, ARL13B, ARL3, ARL6, ARM9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPRG1P1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, XPNPEP3, ZNF423 <b>Nephronophthise (NPHP): 17 Gene (50,3 kb)</b> ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 <b>Bardet-Biedl-Syndrom (BBS): 22 Gene (44,3 kb)</b> ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP <b>Senior-Loken-Syndrom (SLSN): 8 Gene (27,9 kb)</b> CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 <b>Joubert-Syndrom (JBTS): 40 Gene (104,1 kb)</b> AH11, ARL13B, ARL3, ARM9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPRG1P1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423 <b>Meckel-Syndrom (MKS): 14 Gene (36,2 kb)</b> B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPRG1P1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15	4 - 6 Wo	E
<b>Präventionsdiagnostik: Carrier-Screening</b>		
<b>Carrier-Screening *</b> 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, AGR1, AGXT, AH11, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMT, ANK1, ANO10, ANTXR2, APTX, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6VOA2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRIP1, BSND, BTD, BTK, CA2, CAPN3, CASR, CBS, CC2D2A, CCDC88C, CD3D, CD3E, CD40LG, CDH23, CEP290, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNA1, 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, CYP19A1, CYP19A1, CYP11B1, CYP27A1, CYP27B1, D2HGDH, DBT, DCLRE1C, DCX, DDB2, DDC, DDUOX, DHCR24, DHCR7, DHDDS, DKC1, DLD, DLL3, DMD, DMP1, DNAH5, DNAI1, DNAI2, DNAJC19, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPYD, DSP, DUOX2, DUOX2A2, 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, FKBP, FKTN, FLNA, FMO3, FOLR1, FOXN1, FOXP3, FRAS1, FREM2, FUCA1, FXN, G6PC1, G6PC3, G6PD, GAA, GALT, GALE, GALK1, GALNS, GALT, GAMT, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDF5, GFM1, GJA1, GJB1, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNE, GNPTAB, GNPTG, GNS, GP1BA, GP9, GRHRP, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HESX1, HEXA, HEXB, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HOGA1, HPD, HPR1, HPS1, HPS3, HPS4, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPG2, HYAL1, HYL1, 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, MFSDB, MGAT2, MID1, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPI, MPL, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTPP, MUTYH, MVK, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFAF5, NDUFS6, NEU1, NEUROG3, NHLRC1, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NROB1, NR2E3, NTRK1, NUP62, OAT, OCA2, 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, RPRG1P1L, 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, RPRG1P1L, 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, UQCRL, UQCRL, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WAS, WNT10A, WNT3, WNT7A, WRN, WT1, XPA, XPC, ZIC3, ZMPSTE24, ZNF469	4 - 6 Wo	E

Erkrankung/Diagnostik	Dauer	Material
Pränatale Diagnostik: Fetale Anomalien		
<p><b>Fetale Anomalien *</b></p> <p>Gen-Panel: ID850.00, 1223 Gene (3235,6 kb)</p> <p>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, AH11, 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, ARFGF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6VOA2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCL11A, BCOR, BCS1L, BFSP2, BGN, BHLHA9, BICD2, BIN1, BLM, BLTP1, BMP1, BMP2, BMP4, 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, CRIP1, 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, DNML1, DNMT2, DNMT3A, DNMT3B, DOCK6, DOK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSP, DSTYK, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC211, DYNC212, DYNC2L11, DYNLT2B, DYRK1A, DZIP1L, EBF3, EBP, ECEL1, EDA, EDNRA, EDNRB, EED, EFNB1, EFTUD2, EHMT1, EIF2AK3, EIF2B3, EIF2B3, 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, FANGC, FANCI, FANCL, FAR1, FAT4, FBLN5, FBN1, FBN2, FBXL4, FGD1, FGF10, FGF3, FGF8, FGF9, FGF11, FGF12, FGF13, FGF14, FGF15, FGF16, FGF17, FGF18, FGF19, FGF20, FGF21, FGF22, FGF23, FGF24, FGF25, FGF26, FGF27, FGF28, FGF29, FGF30, FGF31, FGF32, FGF33, FGF34, FGF35, FGF36, FGF37, FGF38, FGF39, FGF40, FGF41, FGF42, FGF43, FGF44, FGF45, FGF46, FGF47, FGF48, FGF49, FGF50, FGF51, FGF52, FGF53, FGF54, FGF55, FGF56, FGF57, FGF58, FGF59, FGF60, FGF61, FGF62, FGF63, FGF64, FGF65, FGF66, FGF67, FGF68, FGF69, FGF70, FGF71, FGF72, FGF73, FGF74, FGF75, FGF76, FGF77, FGF78, FGF79, FGF80, FGF81, FGF82, FGF83, FGF84, FGF85, FGF86, FGF87, FGF88, FGF89, FGF90, FGF91, FGF92, FGF93, FGF94, FGF95, FGF96, FGF97, FGF98, FGF99, FGF100, FGF101, FGF102, FGF103, FGF104, FGF105, FGF106, FGF107, FGF108, FGF109, FGF110, FGF111, FGF112, FGF113, FGF114, FGF115, FGF116, FGF117, FGF118, FGF119, FGF120, FGF121, FGF122, FGF123, FGF124, FGF125, FGF126, FGF127, FGF128, FGF129, FGF130, FGF131, FGF132, FGF133, FGF134, FGF135, FGF136, FGF137, FGF138, FGF139, FGF140, FGF141, FGF142, FGF143, FGF144, FGF145, FGF146, FGF147, FGF148, FGF149, FGF150, FGF151, FGF152, FGF153, FGF154, FGF155, FGF156, FGF157, FGF158, FGF159, FGF160, FGF161, FGF162, FGF163, FGF164, FGF165, FGF166, FGF167, FGF168, FGF169, FGF170, FGF171, FGF172, FGF173, FGF174, FGF175, FGF176, FGF177, FGF178, FGF179, FGF180, FGF181, FGF182, FGF183, FGF184, FGF185, FGF186, FGF187, FGF188, FGF189, FGF190, FGF191, FGF192, FGF193, FGF194, FGF195, FGF196, FGF197, FGF198, FGF199, FGF200, FGF201, FGF202, FGF203, FGF204, FGF205, FGF206, FGF207, FGF208, FGF209, FGF210, FGF211, FGF212, FGF213, FGF214, FGF215, FGF216, FGF217, FGF218, FGF219, FGF220, FGF221, FGF222, FGF223, FGF224, FGF225, FGF226, FGF227, FGF228, FGF229, FGF230, FGF231, FGF232, FGF233, FGF234, FGF235, FGF236, FGF237, FGF238, FGF239, FGF240, FGF241, FGF242, FGF243, FGF244, FGF245, FGF246, FGF247, FGF248, FGF249, FGF250, FGF251, FGF252, FGF253, FGF254, FGF255, FGF256, FGF257, FGF258, FGF259, FGF260, 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Erkrankung/Diagnostik	Dauer	Material
<b>Whole Exome Sequencing (WES)</b>		
<b>Analysen</b>		
<b>WES-Solo-Exom *</b> (Index-Patient)	1 - 3 Mo	E
<b>WES-Trio-Exom *</b> (Index-Patient und Eltern)	1 - 3 Mo	E
<b>Hinweise</b>		
* = akkreditiertes Verfahren <b>HG Mannheim</b> und <b>HG München</b>		
# = akkreditiertes Verfahren <b>HG Freiburg</b>		
<b>E</b> = EDTA-Blut, 3 - 5 ml		