

Neuromuskuläre Erkrankungen und Neurodegenerative Erkrankungen			
Panel-ID			Bearbeitungszeit
ID209	Amyotrophe Lateralsklerose (ALS)	Amyotrophe Lateralsklerose (ALS): 31 Gene (71,4 kb) ALS2, ANG, ANXA11, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP Amyotrophe Lateralsklerose und Frontotemporale Demenz (FTDALS): 13 Gene (21,2 kb) CCNF, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Amyotrophe Lateralsklerose, juvenile Form: 4 Gene (21,0 kb) ALS2, SETX, SIGMAR1, SPG11	3 - 5 Wochen
ID086	Sensorisch-autonome Neuropathie (HSAN, HSN)	Sensorisch-autonome Neuropathie (HSAN, HSN): 15 Gene (52,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, WNK1	3 - 5 Wochen
ID052	Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT, HMSN)	Charcot-Marie-Tooth-Neuropathie, axonale Form (CMT, HMSN): 41 Gene (98,5 kb) AARS1, AIFM1, ATP1A1, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PRPS1, RAB7A, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth-Neuropathie, axonal, autosomal-dominant (CMT2): 22 Gene (60,2 kb) AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, TRPV4, VCP Charcot-Marie-Tooth-Neuropathie, axonal, autosomal-rezessiv (CMT2): 10 Gene (25,1 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MME, MPV17, SPG11, TRIM2 Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth-Neuropathie, X-chromosomal (CMTX): 4 Gene (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Charcot-Marie-Tooth-Neuropathie mit Optikusatrophie (CMT6): 3 Gene (4,5 kb) MFN2, PDXK, SLC25A46	4 - 6 Wochen

Panel-ID			Bearbeitungszeit
ID051	Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT, HMSN)	Charcot-Marie-Tooth-Neuropathie, demyelinisierende Form (CMT, HMNS): 26 Gene (56,7 kb) CNTNAP1, COX6A1, DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-dominant (CMT1): 7 Gene (6,3 kb) EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22 Charcot-Marie-Tooth-Neuropathie, demyelinisierend, autosomal-rezessiv (CMT4): 12 Gene (33,8 kb) EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth-Neuropathie, intermediär (CMTDI, CMTRI): 11 Gene (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Hypertrophe Déjerine-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 Wochen
ID254	Distale motorische Neuropathie (HMN, DSMA)	Distale motorische Neuronopathie (HMN, DSMA): 17 Gene (38,7 kb) ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4, WARS1 Distale motorische Neuronopathie, dominant (HMN, DHMN): 12 Gene (26,7 kb) BSCL2, DCTN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, TRPV4, WARS1 Distale motorische Neuronopathie, rezessiv (DSMA): 5 Gene (12,0 kb) ATP7A, DNAJB2, IGHMBP2, PLEGHG5, SIGMAR1	3 - 5 Wochen
ID195	Essentieller Tremor (ETM)	Essentieller Tremor (ETM): 4 Gene (16,6 kb) DRD3, FUS, SCN4A, TENM4	3 - 5 Wochen
ID216	Hyperekplexie (HKPX)	Hyperekplexie (HKPX): 9 Gene (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 Wochen
ID255	Myotonie	Myotonie: 5 Gene (25,0 kb) ATP2A1, CLCN1, HINT1, HSPG2, SCN4A Ggf. DMPK- und CNBP-Repeat-Analyse (DM1, DM2)	3 - 5 Wochen

Panel-ID			Bearbeitungszeit
ID253	Periodische Paralyse	Periodische Paralyse: 7 Gene (16,6 kb) CACNA1S, KCNE3, KCNJ2, KCNJ5, KCNJ12, KCNJ18, SCN4A	3 - 5 Wochen
ID152	Spinale Muskelatrophie (SMA)	Spinale Muskelatrophie (SMA): 31 Gene (69,5 kb) ASA1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS1 Proximale spinale Muskelatrophie (SMA): 10 Gene (26,6 kb) ASA1, ASCC1, BICD2, CHCHD10, DYNC1H1, SMN1, SMN2, TRIP4, UBA1, VAPB Distale spinale Muskelatrophie (DSMA, HMN): 18 Gene (41,9 kb) ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, SIGMAR1, PLEKHG5, REEP1, SETX, SLC5A7, TRPV4, UBA1, WARS1 Ggf. AR-Repeat-Analyse (SMAX1)	3 - 5 Wochen
ID148	Spastische Paraplegie (SPG, HSP)	Spastische Paraplegie (SPG, HSP): 58 Gene (117,9 kb) ALDH18A1, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, L1CAM, MAG, MTRFR, NIPA1, NT5C2, PCYT2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SELENO1, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, TECPR2, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27 Spastische Paraplegie, autosomal-dominant: 17 Gene (33,9 kb) ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, UBAP1, WASHC5, ZFYVE27 Spastische Paraplegie, autosomal-rezessiv: 44 Gene (92,3 kb) AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NT5C2, PCYT2, PLP1, PNPLA6, REEP2, SELENO1, SPART, SPG7, SPG11, SPG21, TECPR2, TFG, UCHL1, VPS37A, ZFYVE26 Spastische Paraplegie, X-chromosomal: 2 Gene (4,6 kb) L1CAM, PLP1	4 - 6 Wochen
ID228	Spastische Ataxie (SPAX)	Spastische Ataxie (SPAX): 12 Gene (34,8 kb) AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1	3 - 5 Wochen

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ID276	Hereditäre Ataxien, umfassende Diagnostik	<p>Hereditäre Ataxien, umfassende Diagnostik: 114 Gene (334,2 kb)</p> <p>ABCB7, ABHD12, ACO2, AFG3L2, AIFM1, ANO10, APTX, ATCAY, ATG5, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CASK, CCDC88C, CHP1, CLCN2, CLN5, COA7, COQ8A, CWF19L1, CYP27A1, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, FLVCR1, GDAP2, GOSR2, GRID2, GRM1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, LAMA1, MARS2, MME, MRE11, MTCL1, MTPAP, MTTP, NBN, NKX6-2, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PLD3, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRKCG, PRPS1, PTF1A, PUM1, RNF216, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1</p> <p>Episodische Ataxie (EA): 6 Gene (20,5 kb)</p> <p>ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3</p> <p>Spastische Ataxie (SPAX): 6 Gene (24,7 kb)</p> <p>AFG3L2, CHP1, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1</p> <p>Spinozerebelläre Ataxie, autosomal-dominant (SCA): 25 Gene (85,6 kb)</p> <p>AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4*, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, PDYN, PLD3, PRKCG, PUM1, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2</p> <p>Spinozerebelläre Ataxie, autosomal-rezessiv (SCAR): 24 Gene (87,8 kb)</p> <p>ANO10, ATG5, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PMPCA, RUBCN, SCYL1, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP2, THG1L, TPP1, UBA5, VPS13D, VWA3B, WWOX, XRCC1</p> <p>Spinozerebelläre Ataxie mit axonaler Neuropathie (SCAN): 3 Gene (10,6 kb)</p> <p>COA7, SETX, TDP1</p> <p>Zerebelläre Ataxie mit mentaler Retardierung (CAMRQ): 4 Gene (12,9 kb)</p> <p>ATP8A2, CA8, VLDLR, WDR81</p> <p>Zerebelläre Ataxie, X-chromosomal: 7 Gene (15,9 kb)</p> <p>ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6</p> <p>Ataxie mit okulomotorischer Apraxie (AOA): 4 Gene (13,3 kb)</p> <p>APTX, PIK3R5, PNKP, SETX</p> <p>Ataxia teleangiectatica (AT): 5 Gene (15,4 kb)</p> <p>APTX, ATM, MRE11, NBN, PCNA</p>	4 - 6 Wochen

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ID184	Episodische Ataxie (EA)	Episodische Ataxie (EA): 6 Gene (20,6 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3	3 - 5 Wochen
ID213	Zerebelläre Ataxie, autosomal-rezessiv	Zerebelläre Ataxie, autosomal-rezessiv: 50 Gene (164,7 kb) ANO10, APTX, ATCAY, ATG5, ATM, ATP8A2, CA8, COA7, COQ8A, CWF19L1, CYP27A1, FXN, GDAP2, GRID2, GRM1, PEX7, PHYH, PIK3R5, PMPCA, PNKP, PNPLA6, POLG, RNF216, RUBCN, SCYL1, SACS, SETX, SIL1, SLC52A2, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TPPA, TWNK, UBA5, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1 Spinozerebelläre Ataxie (SCAR): 24 Gene (87,8 kb) ANO10, ATG5, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PMPCA, RUBCN, SCYL1, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP2, THG1L, TPP1, UBA5, VPS13D, VWA3B, WWOX, XRCC1 Spinozerebelläre Ataxie mit axonaler Neuropathie (SCAN): 3 Gene (10,6 kb) COA7, SETX, TDP1 Zerebelläre Ataxie mit okulomotorischer Apraxie (AOA): 4 Gene (13,3 kb) APTX, PIK3R5, PNKP, SETX Zerebelläre Ataxie mit mentaler Retardierung (CAMRQ): 4 Gene (12,9 kb) ATP8A2, CA8, VLDLR, WDR81	4 - 6 Wochen
ID236	Zerebelläre Ataxie, autosomal-dominant	Zerebelläre Ataxie, autosomal-dominant: 29 Gene (95,6 kb) AFG3L2, ATP1A3, CACNA1A, CACNA1G, CCDC88C, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, PDYN, PLD3, PRKCG, PUM1, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2, TUBB4A Ggf. SCA1-, SCA2-, SCA3-, SCA6-, SCA7-Repeat-Analyse	3 - 5 Wochen
ID273	Zerebelläre Ataxie, X-chromosomal	Zerebelläre Ataxie, X-chromosomal: 7 Gene (15,9 kb) ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6	3 - 5 Wochen
ID077	Parkinson-Krankheit (PARK)	Parkinson-Krankheit (PARK): 21 Gene (57,6 kb) ADH1C, ATP13A2, CHCHD2, DNAJC6, EIF4G1, FBXO7, GBA, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, PRKN, SNCA, SYNJ1, UCHL1, VPS13C, VPS35 Parkinson-Krankheit, early-onset: 9 Gene (30,3 kb) ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, SYNJ1, VPS13C Parkinson-Krankheit, late-onset: 12 Gene (27,3 kb) ADH1C, CHCHD2, EIF4G1, GBA, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, SNCA, UCHL1, VPS35	3 - 5 Wochen

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ID128	Dystonie (DYT)	<p>Dystonie (DYT): 23 Gene (49,7 kb) ANO3, ACTB, ATP1A3, COL6A3, ECHS1, GCH1, GNAL, HPCA, KCTD17, KMT2B, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VPS16</p> <p>Dystonie (DYT), isolierte Form: 8 Gene (25,5 kb) ANO3, COL6A3, GNAL, HPCA, KMT2B, THAP1, TOR1A, TUBB4A</p> <p>Dystonie (DYT), kombinierte Form: 15 Gene (24,4 kb) ACTB, ATP1A3, ECHS1, GCH1, KCTD17, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, VPS16</p>	3 - 5 Wochen
ID286	Paroxysmale Dyskinesie	<p>Paroxysmale Dyskinesie: 14 Gene (38,9 kb) ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24</p>	3 - 5 Wochen
ID272	Choreatiforme Bewegungsstörung	<p>Choreatiforme Bewegungsstörung: 17 Gene (40,6 kb) ADCY5, ATP1A2, ATP1A3, FRRS1L, GNAO1, HPRT1, KCNMA1, NKX2-1, PDE2A, PDE10A, PNKD, PRNP, PRRT2, RNF216, SLC2A1, VPS13A, XK</p>	3 - 5 Wochen
ID200	Arthrogrypose	<p>Arthrogrypose: 61 Gene (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNM2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNT2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2</p> <p>Arthrogryposis multiplex congenita (AMC): 6 Gene (58,2 kb) ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1</p> <p>Distale Arthrogrypose (DA): 11 Gene (40,3 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNT2, TNNT3, TPM2, UBA1</p> <p>Letales kongenitales Kontraktursyndrom (LCCS): 11 Gene (31,5 kb) ADCY6, ADGRG6, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42</p> <p>Fetale Akinesie-Deformation-Sequenz (FADS): 4 Gene (7,6 kb) DOK7, MUSK, NUP88, RAPSN</p>	4 - 6 Wochen

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ID196	Distale Arthrogrypose (DA)	Distale Arthrogrypose (DA): 10 Gene (39,8 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, PIEZO2, TNNI2, TNNT3, TPM2, UBA1	3 - 5 Wochen
ID130	Kongenitales myasthenes Syndrom (CMS)	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, präsynaptisch: 8 Gene (21,9 kb) AGRN, CHAT, MYO9A, SLC5A7, SLC18A3, SLC25A1, SYT2, VAMP1 Kongenitales myasthenes Syndrom, 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 Wochen
ID178	Walker-Warburg-Syndrom (WWS, MDDGA)	Walker-Warburg-Syndrom (WWS, MDDGA): 14 Gene (23,7 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wochen
ID122	Gliedergürtelmuskeldystrophie (LGMD, MDDGC)	Gliedergürtelmuskeldystrophie (LGMD, MDDGC): 35 Gene (191,7 kb) ANOS5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DNAJB6, DPM3, DYSF, FKRP, FKTN, GMPPB, HNRNPDL, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPP11, TRIM32, TTN Gliedergürtelmuskeldystrophie, autosomal-rezessiv (LGMDR): 31 Gene (184,2 kb) ANOS5, BVES, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DPM3, DYSF, FKRP, FKTN, GMPPB, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TRAPP11, TRIM32, TTN Gliedergürtelmuskeldystrophie, autosomal-dominant (LGMDD): 7 Gene (23,2 kb) CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3 Gliedergürtelmuskeldystrophie-Dystroglykanopathie (MDDGC): 11 Gene (17,6 kb) CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2 Ullrich-Muskeldystrophie (UCMD): 3 Gene (15,7 kb) COL6A1, COL6A2, COL6A3 Bethlem-Myopathie (BTHLM): 3 Gene (15,7 kb) COL6A1, COL6A2, COL6A3	4 - 6 Wochen

Panel-ID			Bearbeitungszeit
ID179	Muskeldystrophie-Dystroglykanopathie (MDDGA, MDDGB, MDDGC)	<p>Muskeldystrophie-Dystroglykanopathie (MDDG): 15 Gene (24,0 kb) B3GALNT2, B4GAT1, DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1</p> <p>Muskeldystrophie-Dystroglykanopathie (MDDGA) mit Hirn- und Augenomalien: 14 Gene (22,8 kb) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD*, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1</p> <p>Muskeldystrophie-Dystroglykanopathie (MDDGB) mit oder ohne intellektuelle Entwicklungsstörung: 8 Gene (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2</p> <p>Muskeldystrophie-Dystroglykanopathie (MDDGC), Gliedergürtelmuskeldystrophie: 11 Gene (18,5 kb) DAG1, DPM3, FKRP, FKTN, GMPPB, ISPD, POMGNT1, POMGNT2, POMK, POMT1, POMT2</p>	3 - 5 Wochen
ID212	Kongenitale Strukturmyopathie	<p>Kongenitale Strukturmyopathie: 20 Gene (78,8 kb) ACTA1, BIN1, CCDC78, CFL2, DNM2, KBTBD13, KLHL40, KLHL41, LMOD2, MAP3K20, MTM1, MTMR14, MYPN, NEB, RYR1, SELENON, SPEG, TNNT1, TPM2, TPM3</p> <p>Core-Myopathie: 4 Gene (18,9 kb) ACTA1, RYR1, SELENON, TPM3</p> <p>Nemalin-Myopathie (NEM): 11 Gene (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, TNNT1, TPM2, TPM3, MYPN, NEB</p> <p>Zentronukleäre Myopathie (CNM): 7 Gene (21,4 kb) CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG</p>	3 - 5 Wochen
ID161	Myofibrilläre Myopathie (MFM)	Myofibrilläre Myopathie (MFM): 9 Gene (119,3 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYOT, PYROXD1, TTN	4 - 6 Wochen
ID121	Emery-Dreifuss-Muskeldystrophie (EDMD)	Emery-Dreifuss-Muskeldystrophie (EDMD): 6 Gene (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43	3 - 5 Wochen
ID264	Neurodegeneration mit Eisenablagerung im Gehirn (NBIA)	Neurodegeneration mit Eisenablagerung im Gehirn (NBIA): 11 Gene (20,0 kb) ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45	3 - 5 Wochen

ID277	Hypomyelinisierende Leukodystrophie (HLD)	<p>Hypomyelinisierende Leukodystrophie (HLD): 22 Gene (35,0 kb)</p> <p>AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11</p>	3 - 5 Wochen
ID204	Leukodystrophie und Leukoenzephalopathie, umfassende Diagnostik	<p>Leukodystrophie und Leukoenzephalopathien, umfassende Diagnostik: 123 Gene (208,5 kb)</p> <p>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, SNORD11B, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYMP, TYROBP, UFM1, VPS11, ZFYVE26</p> <p>Leukodystrophie mit Hypomyelinisierung (HLD): 22 Gene (35,0 kb)</p> <p>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)</p> <p>PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26</p> <p>Orthochromatische Leukodystrophie: 10 Gene (14,6 kb)</p> <p>ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1</p> <p>Metachromatische Leukodystrophie: 3 Gene (4,2 kb)</p> <p>ARSA, PSAP, SUMF1</p> <p>Aicardi-Goutières-Syndrom (AGS): 7 Gene (11,9 kb)</p> <p>ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1</p> <p>CADASIL, CARASIL: 2 Gene (8,4 kb)</p> <p>HTRA1, NOTCH3</p>	4 - 6 Wochen

Kosten

Die Kosten werden bei bestehender medizinischer Indikation über einen Überweisungsschein Typ 10 (EBM) abgerechnet. Humangenetische Leistungen sind nicht budgetrelevant. Für privatversicherte Patienten sowie private Kostenträger (Krankenhäuser etc.) können auf Wunsch entsprechende Kostenvoranschläge erstellt werden.