



Einsender:

Patientendaten:

Rechnung: Überweisungsschein (Nr. 10) privat stationär ambulant

Untersuchungsanforderung Neurologische Erkrankungen / Muskuläre Erkrankungen

Untersuchungsgrund: diagnostisch prädiktiv auf familiäre Mutation _____

Klinische Angaben:

Familienanamnese:

- Patient selbst klinisch betroffen
- es liegen keine molekulargenetischen Voruntersuchungen vor
- folgende molekulargenetische Vorbefunde wurden erhoben: _____

Entnahmedatum / Uhrzeit:

Unterschrift:

Bitte stellen Sie für die Versendung folgende Dokumente und Proben zusammen:

- Einwilligungserklärung des Patienten gemäß GenDG und Untersuchungsanforderung
- Überweisungsschein Nr. 10 (für Kassenpatienten) bzw. Kostenübernahmeerklärung (für Privatpatienten)
- 5 ml EDTA-Blutröhrchen mit Patientennamen und Geburtsdatum beschriftet

Patientenname: _____

Geb.datum: _____

Neurologische Erkrankungen

- Amyotrophe Lateralsklerose**
ALS2, ANG, CHCHD10, FUS, MATR3, SETX, SOD1, TARDBP, VAPB, CHMP28, UBQLN2, VCP
- Ataxie**
AAAS, ABCB7, ABHD12, AFG3L2, AMPD2, ANO10, AP1S2, APTX, ARSA, ATCAY, ATM, ATP1A3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CCDC88C, CLCN2, CLN6, COA7, COQ8A, COX20, CP, CTBP1, CWF19L1, CYP27A1, CYP2U1, DARS2, DDHD2, DNAJC19, DNAJC5, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EPM2A, FGF14, FLVCR1, FOLR1, FXN, GBA2, GJC2, GOSR2, GPAA1, GRID2, GRM1, HEXB, HSD17B4, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, MARS2, MRE11, MTPP, NHLRC1, NKX6-2, NPC1, NPC2, OPA3, OPHN1, PDYN, PEX16, PEX7, PLA2G6, PMPCA, PNKP, PNPLA6, POLG, POLR3A, PRICKLE1, PRKCG, PRNP, PUM1, RNF170, RNF216, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC2A1, SLC9A6, SNX14, SOD1, SPG7, SPTBN2, STUB1, SYNE1, TDP1, TDP2, TGM6, TMEM240, TPP1, TTBK2, TTC19, TTPA, TWNK, VAMP1, VLDLR, VPS13D, WDR81, WFS1, WWOX, XRCC1
- Demenz**
APOE, APP, CHCHD10, CHMP2B, CSF1R, FUS, GRN, ITM2B, MAPT, NOTCH3, PRNP, PSEN1, PSEN2, SQSTM1, TARDBP, TBK1, TREM2, UBQLN2, VCP
- Dystonie**
ACY1, ADAR, ADCY5, ANO3, APTX, ARX, ATM, ATP13A2, ATP1A2, ATP1A3, ATP7B, BCAP31, C19orf12, CACNA1B, CIZ1, COASY, COL6A3, DCAF17, DLAT, DNAJC12, FA2H, FBXO7, FRRS1L, FTL, GCDH, GCH1, GNAL, GNAO1, GNB1, GPR88, GRIN1, HPCA, HTRA2, IRF2BPL, KCNMA1, KCTD17, KMT2B, MECR, NKX2-1, NKX6-2, PANK2, PARK7, RELN, PDE2A, PDHA1, PDHX, PINK1, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, SCN8A, SERAC1, SGCE, SLC19A3, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SPR, STXBP1, SYNJ1, SYT1, TAF1, TBCD, TH, THAP1, TOR1A, TTPA, TUBB4A, UBTF, UNC13A, VAC14, VPS13A, WDR45, YY1
- Hereditäre Neuropathien**
AARS1, ABCA1, ABHD12, AIFM1, ALS2, ANG, APTX, ARHGEF10, ASAH1, ASCC1, ATLL1, ATLL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, C12orf65, CCT5, CHCHD10, CLP1, CNTNAP1, COA7, COX10, COX6A1, CTDP1, CYP27A1, DCAF8, DCTN1, DGAT2, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EXOSC3, EXOSC8, FAM126A, FAM134B, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FUS, FXN, GALC, GAN, GARS1, GDAP1, GJB1, GLA, GMPPA, GNB4, GSN, HADHA, HADHB, HARS1, HINT1, HK1, HOXD10, HSBP3, HSPB1, HSPB8, IFRD1, IGHMBP2, IKBKAP, INF2, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MED25, MFN2, MICAL1, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NDUFAF5, NEFH, NEFL, NGF, NIPA1, NTRK1, OPA1, OPTN, PDHA1, PDK3, PDXK, PEX12, PEX7, PHYH, PLEKHG5, PLP1, PMP2, PMP22, PNKP, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, RNF170, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SEPTIN9, SGPL1, SH3BP4, SH3TC2, SLC12A6, SLC25A46, SLC5A7, SORD, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, SYT2, TDP1, TECPR2, TFG, TRIM2, TRPA1, TRPV4, TTR, TUBB3, TWNK, TYMP, VCP, WNK1, YARS1, ZFH2, HMBS, SCO2, SETX, SIGMAR1, TRIP4, VAPB, UBA1, VRK1
- Hereditäre spastische Spinalparalyse**
AAAS, ABCD1, ADAR, AFG3L2, AIMP1, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ARSA, ATL1, ATP7B, B4GALNT1, BICD2, BSCL2, BTBD9, C12orf65, C19orf12, CAPN1, CCT5, CLCN2, CPT1C, CYP2U1, CYP7B1, DARS2, DDHD1, DDHD2, DNAJC12, DNM2, DSTYK, EIF2B5, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAM134B, FARS2, FRR1L, GAD1, GALC, GAN, GBA2, GBE1, GCH1, GJC2, GNAO1, GNB1, GRID2, GRP88, HSPD1, IBA57, IFIH1, KDM5C, KIAA0196, KIDINS220, KIF1A, KIF1C, KIF5A, KLC4, KMT2B, L1CAM, LYST, MAG, MARS, MARS2, MMACHC, MTHFR, NIPA1, NKX6-2, NT5C2, OPA3, PANK2, PGAP1, PLA2G6, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RNASEH2B, RTN2, SACS, SETX, SLC16A2, SLC33A1, ALC39A14, SOD1, SPART, SPAST, SPG11, SPG20, SPG21, SPG7, SPR, SYNE1, TBCD, TECPR2, TFG, TH, TTR, TUBB4A, UBAP1, UBQLN2, UBTF, UCHL1, UNC13A, USP8, VAC14, VAMP1, VCP, VPS13D, VPS37A, WASHC5, WWOX, ZFYVE26, ZFYVE27
- Periodische Paralysen**
ATP1A2, CACNA1S, KCNE3, KCNJ2, KCNJ5, SCN4A
- Neurodegeneration mit Eisenablagerungen**
ATP13A2, C19orf12, CP, COASY, DCAF17, FA2H, FBXO7, FTL, PANK2, PLA2G6, WDR45
- Parkinson**
ATP1A3, ATP13A2, C19orf12, CHCHD2, DCTN1, DNAJC6, FTL, GBA, GCH1, GRN, LRRK2, MAPT, PANK2, PARK7, PINK1, PLA2G6, PRKRA, PRKN, SLC30A10, SLC39A14, SLC6A3, SNCA, SPG11, SPR, SYNJ1, TH, VPS13C, VPS35
- Paroxysmale Dyskinesie**
DEPDC5, ECHS1, KCNA1, KCNMA1, NKX2-1, PDHA1, PNKD, PRRT2, SCN8A, SLC2A1

Patientenname: _____ Geb.datum: _____

Muskuläre Erkrankungen

- Gliedergürtelmuskeldystrophie**
ANO5, BVES, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, HNRNPDL, ISPD, LAMA2, LMNA, MYOF, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PYROXD1, SGCA, SGCB, SGCD, SGCG, SYNE1, TCAP, TNPO3, TOR1Aip1, TRAPPC11, TRIM32, TTN, VCP
- Kongenitalemyasthene Syndrome**
ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, AGL, ALDOA, AMPD1, CPT1A, ENO3, ETFA, ETFB, ETFDH, FLAD1, G6PC, GAA, GBE1, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, NPL, PDHA1, PFKM, PGK, PGM1, PGAM2, PHKA1, PHKB, PHKG2, POLG2, PNPLA2, PRKAG2, PRM2B, PUS1, PYGM, RBCK1, SLC16A1, SLC22A5, SLC25A20, TAZ, YARS2,
- Kongentiale und distale Myopathien**
ABCC9, ACTA1, ACTN2, ANO5, BAG3, BIN1, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, COL6A3, CNTN1, COL12A1, COL6A1, COL6A2, COX6A2, CRYAB, DES, DNA2, DNAJB5, DNAJB6, DNM2, DYSF, FHL1, FKBP14, FLNC, FXR1, GNE, HACD1, HSPB8, KBTBD13, KLHL9, ISCU, KLHL40, KLHL41, KY, LAMP2, LDB3, LMOD3, LRP12, MAP3K20, MATR3, MB, MEGF10, MICU1, MSTO1, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYPN, MYOD1, MYOT, NEB, ORAI1, PAX7, POLG, POLG2, PUS1, PYROXD1, RRM2B, RYR1, RYR3, SCN4A, SELENON, SIL1, SLC25A42, SLC25A21, SOD1, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, TIA1, TIMM22, TK2, TMEM65, TNNT1, TOR1AIP1, TPM2, TPM3, TRIM32, TRIM54, TRIM63, TTN, TWNK, VCP, VMA21, YARS2
- Metabolische Myopathie**
ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, AGL, ALDOA, AMPD1, CPT1A, ENO3, ETFA, ETFB, ETFDH, FLAD1, G6PC, GAA, GBE1, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, NPL, PDHA1, PFKM, PGAM2, PGK, PGM1, PHKA1, PHKB, PHKG2, PNPLA2, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, SLC16A1, SLC22A5, SLC25A20, TAZ, YARS2
- Muskuläre Erkrankungen (Gesamt-Panel)**
*(270 Gene *)*
- Skapuloperoneale Myopathie**
CAPN3, CAV3, DES, FKRP, GAA, MYH7, SGCA, TRPV4, VCP
- Spinale Muskelatrophie**
AARS, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC8, GARS, HEXA, HSPB1, HSPB8, IGHMBP2, LAS1L, PLEKHG5, RBM7, REEP1, SCO2, SETX, SIGMAR1, SLC5A7, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, YARS

* Auf Nachfrage kann eine ausführliche Genliste zur Verfügung gestellt werden.