



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-Blut Röhrchen 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, ATL3, 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, ATLL1, 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.