Neurology

Hereditary Ataxia

AAAS	ABCB7	ABHD12	AFG3L2	AMPD2	ANO10	AP1S2	APTX	ARSA	ATCAY	ATM
ATP1A3	CA8	CACNA1A	CACNA1G	CAMTA1	CASK	CHMP1A	CLN6	COQ8A	COX20	CP
CWF19L1	CYP27A1	CYP2U1	DARS2	DDHD2	DNAJC5	DNMT1	EIF2B1	EIF2B2	EIF2B3	EIF2B4
EIF2B5	ELOVL4	EPM2A	EXOSC3	FGF14	FLVCR1	FOLR1	FXN	GBA2	GJC2	GOSR2
GRID2	GRM1	HEXA	HEXB	ITPR1	KCNA1	KCNC3	KCND3	KCNJ10	KIF1C	MARS2
MMACHC	MRE11	MTTP	NHLRC1	NPC1	NPC2	OPHN1	PAX6	PDYN	PEX16	PLA2G6
PMPCA	PNKP	PNPLA6	POLG	POLR3A	PRKCG	PRNP	PRRT2	RARS2	RNF170	RNF216
SACS	SAR1B	SEPSECS	SETX	SIL1	SLC1A3	SLC2A1	SLC9A6	SNX14	SPG7	SPTBN2
SRD5A3	STUB1	SYNE1	TGM6	TMEM240	TPP1	TSEN2	TSEN54	TTBK2	TTC19	TTPA
TUBB4A	TWNK	VLDLR	VRK1	WDR73	WDR81	WFS1	WWOX			

Hereditary Spastic Paraplegia (Stage 1)

ATL1*	BSCL2	CYP7B1	FA2H	GJC2	HSPD1	KIAA0196	KIF5A	L1CAM	NIPA1	PLP1
REEP1	SLC33A1	SPG7	SPG11	SPG20	SPG21	SPAST*	ZEB2	ZFYVE27		

*MLPA copy number analysis

Hereditary Spastic Paraplegia (Stage 2 – performed if stage 1 is negative)

ABCD1	ADAR	AFG3L2	AIMP1	ALDH18A1	ALS2	AP4B1	AP4E1	AP4M1	AP4S1	ARG1
ATP13A2	B4GALNT1	C12ORF65	C19ORF12	CAPN1	CYP27A1	CYP2U1	DDHD1	DDHD2	ERLIN1	ERLIN2
FARS2	GBA2	HACE1	KIDINS220	KIF1A	NT5C2	OPA3	PNPLA6	POLR3A	RTN2	SACS
SERAC1	SLC16A2	SLC1A4	SLC25A46	SLC2A1	TUBB4A	WDR45B	ZFYVE26			

Rhabdomyolysis AND Metabolic Myopathies

ACADVL	AGL	ALDOA	ANO5	CACNA1S	CAPN3	CAV3	CPT2	DMD	DYSF	ENO3
ETFA	ETFB	ETFDH	FKRP	GAA	GBE1	GMPPB	GYG1	GYS1	HADHA	HADHB
ISCU	LDHA	LPIN1	PFKM	PGAM2	PGK1	PGM1	PHKA1	PNPLA2	PYGM	RBCK1
RYR1	SLC22A5	TANGO2								

Charcot Marie Tooth / Hereditary Motor and Sensory Neuropathies

AARS	ATL1	ATP7A	BICD2	BSCL2	CCT5	DCTN1	DMN2	DMNT1	DYNC1H1	EGR2
FAM134B	FGD4	FIG4	GARS	GDAP1	HINT1	HSPB1	HSPB3	HSPB8	IGHMBP2	IKBKAP
INF2	KIF1A	LITAF	LMNA	LRSAM1	MARS	MTMR2	NDRG1	NEFL	NGF	NTRK1
PLEKHG5	PRPS1	PRX	RAB7A	REEP1	SBF2	SCN9A	SETX	SH3TC2	SLC52A1	SLC52A2
SLC52A3	SORD	SPTLC1	SPTLC2	TRPV4	VCP	WNK1	YARS			

Spinal Muscular Atrophy / Distal Hereditary Motor Neuropathies

AARS	ASAH1	ATP7A	BICD2	BSCL2	CHCHD10	DCTN1	DNAJB2	DYNC1H1	EXOSC3	EXOSC8
FBXO38	FIG4	GARS	HEXA	HSPB1	HSPB3	HSPB8	IGHMBP2	LAS1L	MATR3	MFN2
PLEKHG5	REEP1	SCO2	SETX	SIGMAR1	SLC52A2	SLC52A3	SLC5A7	SOD1	SYT2	TRPV4
UBA1	VAPB	VCP	VRK1							

Brain Iron Accumulation

ATP13A2	C19orf12	COASY	CP	DCAF17	FA2H	FTL	FUCA1	KIF1A	KMT2B	MECR
PANK2	PLA2G6	PSEN1	SCP2	SLC39A14	SQSTM1	TRIM32	UBTF	VPS13A	WDR45	

Arthrogryposis

ACTA1	ADAMTS10	ANTXR2	ASCC1	ASXL1	B3GALNT2	B4GAT1	BICD2	CHAT	CHRNA1	CHRNB1
CHRND	CHRNE	CHRNG	CHST14	CNTNAP1	COL12A1	COL6A1	COL6A2	COL6A3	COLQ	DAG1
DNM2	DOK7	DPAGT1	DYNC1H1	ECEL1	ERCC6	ERCC8	EXOSC3	FAM20C	FBN2	FGFR2
FKBP10	FKRP	FKTN	GBA	GBE1	GLDN	GLE1	GMPPB	ADGRG6	HSPG2	ISPD
KLHL40	KLHL41	LAMA2	LARGE1	LMOD3	MAGEL2	MPZ	MTM1	MUSK	MYBPC1	MYH2
MYH3	MYH7	MYH8	NALCN	NEB	PEX1	PEX10	PEX11B	PEX12	PEX13	PEX14
PEX16	PEX19	PEX2	PEX26	PEX3	PEX5	PEX6	PEX7	PFKM	PIEZO2	PLOD1
PLOD2	POMGNT1	POMGNT2	POMK	POMT1	POMT2	POR	PRG4	RAPSN	RYR1	SCARF2
SCN4A	SKI	SLC5A7	SMAD4	STAC3	SYNE1	TMEM5	TNNI2	TNNT1	TNNT3	TPM2
TPM3	TRPV4	TSEN54	UBA1	VAMP1	VIPAS39	VPS33B	ZC4H2			

Musculoskeletal

Chondrodysplasia Punctata

AGPS	ARSE	EBP	GNPAT	PEX7

Osteopetrosis

AMER1	ANKH	CA2	CLCN7	CTSK	FAM20C	FERMT3	LEMD3	LRP5	OSTM1	PTH1R
RASGRP2	SNX10	SOST	TCIRG1	TGFB1	TNFRSF11A	TNFSF11	TYROBP			

Hereditary Multiple Osteochomdromas / Multiple Exostoses

EXT1	EXT2

Rare Cancers

Gorlin syndrome	
PTCH1	**SUFU**

Rhabdoid tumour	
SMARCA4	**SMARCB1**

Respiratory Disorders

Surfactant Metabolism Dysfunction			
ABCA3	NKX2-1	SFTPB	SFTPC

Primary Ciliary Dyskinesia										
ARMC4	C21ORF59	CCDC39	CCDC40	CCDC65	CCDC103	CCDC114	CCDC151	CCNO	DNAAF1	DNAAF2
DNAAF3	DNAAF5	DNAH5	DNAH11	DNAI1	DNAI2	DNAL1	DRC1	DYX1C1	GAS8	LRRC6
MCIDAS	RPGR	RSPH1	RSPH4A	RSPH9	SPAG1	ZMYND10				