

Supplementary Table S1. List of variants detected in each disease.

Disease	Clinical diagnosis	Gene	Variant/s	Inheritance
Ataxia				
	Early-onset cerebellar ataxia	<i>APTX</i> (NM_175073.3)	c.837G>A p.(Trp279Ter) c.837G>A p.(Trp279Ter)	AR
	Early-onset cerebellar ataxia	<i>COQ8A</i> (NM_020247.5)	c.642_649del p.(Ala215ArgfsTer69) c.642_649del p.(Ala215ArgfsTer69)	AR
	Cerebellar ataxia	<i>KCND3</i> (NM_001378969.1)	c.1130C>T p.(Thr377Met)	AD
	Spastic ataxia	<i>POLR3A</i> (NM_007055.3)	c.1909+22G>A c.2090C>T p.(Ser697Leu)	AR
	Cerebellar ataxia	<i>SPG7</i> (NM_003119.4)	c.233T>A p.(p.Leu78Ter) c.233T>A p.(p.Leu78Ter)	AR
	Early-onset cerebellar ataxia	<i>SPG7</i> (NM_003119.3)	c.1529C>T p.(Ala510Val) c.1529C>T p.(Ala510Val)	AR
	Ataxic syndrome and cognitive deterioration	<i>STUB1</i> (NM_005861.4)	c.823_824del p.(Leu275AspfsTer16)*	AD*/AR
	Ataxic syndrome	<i>TTR</i> (NM_000371.4)	c.424G>A p.(Val142Ile)	AD
Spastic paraplegia				
	Spastic paraplegia	<i>ABCD1</i> (NM_000033.4)	c.1998C>G p.(Tyr666Ter)	XLR
	Spastic paraplegia	<i>ABCD1</i> (NM_000033.4)	c.-34_17del	XLR
	Spastic paraplegia	<i>AP5Z1</i> (NM_014855.2)	c80_83delinsTGCTGTAACTGTAAGTAA p.(Arg27_Ile28insLeuTer) exon 2 and 3 deletion	AR
	Spastic paraplegia	<i>CYP7B1</i> (NM_004820.3)	c.1456C>T p.(Arg486Cys) c.884C>A p.(Ala295Glu)	AR
	Spastic paraplegia	<i>CYP7B1</i> (NM_004820.3)	c.1490dup p.(Leu487PhefsTer11) c.1490dup p.(Leu487PhefsTer11)	AR

	Spastic paraplegia	<i>DDHD1</i> (NM_001160148.1)	c.1999A>G p.(Arg667Gly) c.1999A>G p.(Arg667Gly)	AR
	Spastic paraplegia	<i>DDHD2</i> (NM_015214.2)	c.1973G>C p.(Arg658His) c.1978 G>A p.(Asp660His)	AR
	Spastic paraplegia	<i>KIF5A</i> (NM_004984.2)	c.95C>T p.(Pro32Leu)	AD
	Spastic paraplegia	<i>POLR3A</i> (NM_007055.3)	c.1909+22G>A c.3429+1G>A	AR
	Spastic paraplegia	<i>POLR3A</i> (NM_007055.3)	c.1628A>C p.(Gln543Pro) c.1909+22G>A	AR
	Spastic paraplegia	<i>PSEN1</i> (NM_000021.4)	c.1261A>G p.(Thr421Ala)	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	exon 1 deletion	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1413+3_1413+6del	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1142_1143del p.(Phe381TrpfsTer12)	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1414-2A>C	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1245+1G>C	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1157A>G p.(Asn386Ser)	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1688-3C>G	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1493+2_1493+5	AD
	Spastic paraplegia	<i>SPAST</i> (NM_014946.3)	c.1676G>A p.(Gly559Asp)	AD
	Spastic paraplegia	<i>SPG7</i> (NM_003119.3)	c.987+1dupG exons 11 to 15 deletion (MLPA)	AR
	Spastic paraplegia	<i>SPG7</i> (NM_003119.3)	c.773_774del (p.Val258GlyfsTer30) c.1529C>T (p.Ala510Val)	AR
Dystonia				
	Early onset focal dystonia	<i>ACTB</i> (NM_001101.3)	c.547C>T p.(Arg183Trp)	AD
	Dystonia and spasticity	<i>GCH1</i> (NM_000161.2) <i>AFG3L2</i> (NM_006796.2)	c.671A>G p.(Lys224Arg) c.1847A>G p.(616Cys)	AD AD
	Cervical dystonia	<i>ANO3</i> (NM_031418.4)	c.1528G>A (p.Glu510Lys)	AD

	Bulbar palsy with oromandibular dystonia	<i>AOPEP</i> (NM_001193329.3)	c.1036T>C p.(Trp346Arg) c.1036T>C p.(Trp346Arg)	AR
	Dystonia and writers' cramp	<i>THAP1</i> (NM_018105.2)	c.19G>A p.(Ala7Thr)	AD
	Dystonia	<i>TOR1A</i> (NM_000113.3)	c.907_909del p.(Glu303del)	AD
	Blepharospasm	<i>SLC2A1</i> (NM_006516.3)	c.847C>T p.(Gln283Ter)	AD
Parkinson				
	Parkinson	<i>GBA</i> (NM_000157.4)	c.[754T>A;1093G>A] p.(Glu365Lys) p.(Phe252Ile) c.1279G>A p.(Glu427Lys) ¹	RF
	Parkinson	<i>GBA</i> (NM_000157.4)	c.1342G>C p.(Asp448His) ²	RF
	Parkinson	<i>LDLR</i> (NM_000527.5)	c.1845+1G>C ³	AD/AR
	Parkinson	<i>LRRK2</i> (NM_198578.4)	c.4321C>G p.(Arg1441Gly)	AD
	Parkinson	<i>LRRK2</i> (NM_198578.4)	c.6059T>C p.(Ile2020Thr)	AD
	Parkinson	<i>PARK7</i> (NM_007262.5)	hg38: 1: 7969345-7969404 deletion hg38: 1: 7969345-7969404 deletion	AR
	Parkinson	<i>PRKN</i> (NM_004562.3)	c.155del p.(Asn53MetfsTer29) exon 3 deletion (MLPA)	AR
	Parkinson, generalized dystonia and spasticity	<i>WDR45</i> (NM_00707.4)	c.235+5G>A ⁴	XLD
ID				
	ID and epilepsy	<i>CSF1R</i> (NM_005211.4)	c.2442+1G>A	AD
	ID, DD, microcephaly and hypotonia	<i>NIPBL</i> (NM_)	c.1415A>G p.(His472Arg)	XLD/XLR
	ID and ASD	<i>WAC</i> (NM_016628.4)	c.1280_1281delCTinsGAG p.(Ser427Ter)	AD
	ID and facial dysmorphism	<i>KCNT2</i> (NM_198503.5)	c.569G>A p.(Arg190His)	AD
	ID and hydrocephalus	<i>FGFR1</i> (NM_023110.3)	c.880G>A p.(Glu294Lys)	AD
	ID and facial dysmorphism	<i>SOX5</i> (NM_006940.6)	c.637C>T p.(Arg213Ter)	AD
	ID	<i>POU3F3</i> (NM_006236.3)	c.539_578del p.(His180ProfsTer38)	AD
	ID and facial dysmorphism	<i>POGZ</i> (NM_015100.4)	c.2989C>T p.(Arg997Ter)	AD

	ID, facial dysmorphisms and body dysmorphic disorder	<i>CNOT3</i> (NM_014516.3)	c.169C>T p.(Arg57Trp)	AD
	Dysmorphism and procurement delay	<i>PPP2R5D</i> (NM_006245.4)	c.592G>A p.(Glu198Lys)	AD
	Opitz syndrome	<i>MID1</i> (NM_000381.3)	c.602_605del p.(Val201GlyfsTer11)	XLR
ASD				
	Psychomotor delay, ASD, absence of language and macrocephaly	<i>PPP2R5D</i> (NM_006245.4)	c.598G>A p.(Glu200Lys)	AD
	Angelman syndrome	<i>SHANK3</i> (NM_001372044.2)	c.3874G>T p.(Glu1292Ter)	AD
Epilepsy				
	Dravet syndrome	<i>SCN1A</i> (NM_001165963.4)	c.4300T>A p.(Trp1434Arg)	AD
	Epilepsy and developmental delay	<i>SCN1A</i> (NM_001165963.4)	c.4476+5G>A	AD
	Epilepsy and ID	<i>DYRK1A</i> (NM_001396.5)	c.665-1G>T	AD
	Epilepsy and ID	<i>SYNGAP1</i> (NM_006772.3)	c.333del p.(Lys114SerfsTer20)	AD
	Sotos syndrome	<i>NSD1</i> (NM_022455.5)	c.4411C>T p.(Arg1471Ter)	AD
Other neurological disorders				
	White matter lesions and cerebellar atrophy	<i>PDGFRB</i> (NM_002609.4)	c.2959C>T p.(Arg987Trp)	AD
	Cerebral cavernous malformation	<i>PDCD10</i> (NM_007217.4)	c.575dup p.(Ser193LysfsTer36)	AD
	Cerebral cavernous malformation	<i>CCM2</i> (NM_031443.4)	c.55C>T p.(Arg19Ter)	AD