Supplementary Table 1: The current list of locus symbols for hereditary parkinsonism.<sup>1, 2</sup>

Symbol	Gene locus	Gene	Inheri-	Status and remarks
			tance	
PARK1	4q21-22	SNCA	AD	Confirmed
PARK2	6q25.2-q27	PARK2 encoding Parkin	AR	Confirmed
PARK3	2p13	Unknown	AD	Unconfirmed; May represent a risk factor; Gene not found since first described in 1998
PARK4	4q21-q23	SNCA	AD	Erroneous locus (identical to <i>PARK1</i> )
PARK5	4p13	UCHL1	AD	Unconfirmed (not replicated since described in 1998)
PARK6	1p35-p36	PINK1	AR	Confirmed

PARK7	1p36	PARK7 encoding DJ-1	AR	Confirmed
PARK8	12q12	LRRK2	AD	Confirmed; Variations in <i>LRRK</i> 2 gene include risk-conferring variants and disease- causing mutations.
PARK9	1p36	ATP13A2	AR	Confirmed
PARK10	1p32	Unknown	Risk factor	N/A
PARK11	2q36-27	Unknown	AD	Initially described mutations in the GIGYF2 gene later found also in controls; no replication studies confirmed GIGYF2 as causative of PD
PARK12	Xq21-q25	Unknown	Risk	N/A

			factor	
PARK13	2p12	HTRA2	AD or risk factor	Unconfirmed
PARK14	22q13.1	PLA2G6	AR	Confirmed; The majority of cases do not include parkinsonism
PARK15	22q12-q13	FBX07	AR	Confirmed
PARK16	1q32	Unknown	Risk factor	N/A
PARK17*	4p16	VPS35	AD	Confirmed
PARK18*	6p21.3	EIF4G1	AD	Unconfirmed
PARK19*	1p31.3	DNAJC6	AR	Confirmed
PARK20*	21q22.11	SYNJ1	AR	Confirmed

AD, autosomal dominant; AR, autosomal recessive.

\*Not approved by HGNC

Supplementary Table 2: The list of locus symbols for hereditary dystonias<sup>3</sup>

Symbol	Locus	Gene	Inheri-	Comments
			tance	
DYT1	9q32-q34	TOR1A	AD	Confirmed
DYT2	Missing	Unknown	AR	Unconfirmed;
				Missing locus, cases are being
				lumped on the basis of
				inheritance pattern alone
DYT3	Xq13.1	TAF1?	XR	The pathogenicity of TAF1 gene
				mutations remains unconfirmed.
DYT4	19p13.3	TUBB4a	AD	Unconfirmed; (but found in the
				same family by two independent
				groups)
				Missing locus
DYT5a	14q22.1-	GCH1	AD	Confirmed
	22.2			
DYT5b	11p15.5	TH	AR	Confirmed
	2p14-p12	SPR	AR	Not listed
DYT6	8p11.1	THAP1	AD	Confirmed
DYT7	18p	Unknown	AD	Unconfirmed (not replicated since
				first described in 1996)*
DYT8	2q35	MR1	AD	Confirmed
DYT9	1p31	SLC2A1	AD	Confirmed

DYT10	16p11.2-	Unknown	AD	Confirmed
	q12.1			
DYT11	7q21.3	SGCE	AD	Confirmed
DYT12	19q13.2	ATP1A3	AD	Confirmed;
				Not represented in the 'PARK' list
DYT13	1p36	Unknown	AD	Unconfirmed (not replicated since
				first described in 2001)
DYT14	11p15.5	GCH1	AD	Erroneous locus (identical to
				DYT5a)
DYT15	18p11	Unknown	AD	Unconfirmed (not replicated since
				first described in 2002)
DYT16	2q31.2	PRKRA	AR	Confirmed <sup>4</sup>
DYT17	20p11.22	Unknown	AR	Unconfirmed (not replicated since
	-q13.12			symbol in 2008)
DYT18	1p34.2	SLC2A1	AD	Identical to DYT9
DYT19**	16q	PRRT2	AD	Identical to DYT10
DYT20**	2q	Unknown	AD	Unconfirmed (clinical overlap with
				PNKD1; locus very close to
				DYT8)
DYT21**	2q14.3-	Unknown	AD	Unconfirmed
	q21.3			

DYT22	None	None		Symbol officially accepted by
				HGNC but no associated locus or
				gene recorded.
DYT23	9q34	CIZ1	AD	Unconfirmed
DYT24**	11p14.2	ANO3	AD	Unconfirmed
DYT25**	18p11.21	GNAL	AD	Confirmed

AD, autosomal dominant; AR, autosomal recessive; XR, X-linked recessive

\*Unconfirmed but supported by the description of several patients with an 18p deletion syndrome and co-occurrence of dystonia.

\*\*Not approved by HGNC.

Supplementary Table 3: The current list of episodic ataxias <sup>5</sup>

Symbol	Locus	Gene	Inheri-tance	Status or
				comments
EA-1	12p13	KCNA1	AD	Confirmed
EA-2	19p13	CACNA1A	AD	Confirmed
EA-3	Unconfirmed	Unknown	AD	Linkage to
				1q42
				proposed <sup>6</sup>
EA-4	Unknown	Unknown	AD	
EA-5	2q22-q23	CACNB4*	AD	Unconfirmed
EA-6	5p13	SLC1A3	AD	Confirmed
EA-7	Unconfirmed	Unknown	AD	Candidate
				region 19q13
				identified <sup>7</sup>

Allelic to juvenile myoclonic epilepsy

Supplementary Table 4: Current list of SCA loci and genes<sup>8, 9</sup>

	Locus	Gene	Inheri-	Status or comments
			tance	
SCA1	6p23	ATXN1	AD	Confirmed
SCA2	12q24.1	ATXN2	AD	Confirmed
SCA3	14q21	ATXN3	AD	Confirmed
SCA4	16q22.1	-	AD	Unconfirmed
SCA5	11q13	SPTBN2	AD	Confirmed
SCA6	19p13	CACNA1A	AD	Confirmed
SCA7	3p21.1-p12	ATXN7	AD	Confirmed
SCA8	13q21	ATXN8OS	AD	Confirmed
SCA9	-	-	AD	Unconfirmed, no locus
SCA10	22q13.31	ATXN10	AD	Confirmed
SCA11	15q15.2	TTBK2	AD	Confirmed
SCA12	5q32	PPP2R2B	AD	Confirmed
SCA13	19q13.33	KCNC3	AD	Confirmed

SCA14	19q13.4	PRKCG	AD	Confirmed
SCA15	3p26.1	ITPR1	AD	Confirmed SCA15 and SCA16 are allelic
SCA16	3p26.1	ITPR1	AD	Confirmed SCA15 and SCA16 are allelic
SCA17	6q27	TBP	AD	Confirmed
SCA18	7q22-32	-	AD	Unconfirmed
SCA19	1p21-q21	KCND3	AD	Confirmed SCA19 and SCA22 are allelic
SCA20	11q12		AD	Unconfirmed
SCA21	7p21.3-p15.1	TMEM240	AD	Confirmed
SCA22	1p21-q21	KCND3	AD	Confirmed SCA19 and SCA22 are allelic
SCA23	20p13	PDYN	AD	Confirmed

SCA24	1p36	-	AR	Autosomal recessive
				(SCAR4)
SCA25	2p21-p13	-	AD	Unconfirmed
SCA26	19p13.3	eEF2	AD	Unconfirmed
SCA27	13q34	FGF14	AD	Confirmed
SCA28	18p11	AFG3L2	AD	Confirmed
SCA29	3p26	ITPR1	AD	Congenital ataxia
SCA30	4q34.3-q35.1	-	AD	Unconfirmed
SCA31	16q21	BEAN1	AD	Confirmed Previously referred to as SCA4
SCA32	7q32-33	-	AD	Unconfirmed
SCA34	6p12.3-q16.2	-	AD	Unconfirmed
SCA35	20p13	TGM6	AD	Confirmed
SCA36	20p13	NOP56	AD	Confirmed
SCA37	1p32	-	AD	Unconfirmed
SCA38	6р	ELOVL5	AD	Confirmed <sup>10</sup>

SCA40	14q32	CCDC88C	AD	Unconfirmed <sup>11</sup>

Supplementary Table 5: The current list of HD/HDL loci and genes<sup>12</sup>

Locus Symbol	Locus	Gene	Inheritance	Status and remarks
Huntington's disease (HD)	4p16.3	HTT	AD	Confirmed
HDL 1	20.p13	PRNP	AD	Confirmed
HDL 2	16q24.2	JPH3	AD	Confirmed
HDL 3	4p15.3	Unknown	AR	No known gene
HDL 4	6q27	TBD	AD	Confirmed. Also known as SCA17;

Supplementary Table 6: Monogenic forms of hereditary spastic paraplegias (HSPs) <sup>13,</sup>

Symbol	Gene locus	Inheri-	Gene	status and remarks
		tance		
SPG1	Xq28	XR	L1CAM	confirmed
SPG2	Xq21	XR	PLP1	confirmed
SPG3A	14q11-q21	AD/AR	ATL1	confirmed
SPG4	2p22	AD	SPAST	confirmed
SPG5A	8q12-q13	AR	CYP7B1	confirmed
SPG5B	Linkage to chromosom e8	AR	Unknown	unconfirmed
SPG6	15q11.1	AD	NIPA1	confirmed
SPG7	16q24.3	AR/AD	SPG7	confirmed
SPG8	8q24.13	AD	KIAA0196	confirmed

SPG9	10q23.3-	AD	Unknown	confirmed
	24.1			
SPG10	12q13.3	AD	KIF5A	confirmed
SPG11	15a21	AR	KIAA1840	confirmed
	. • 4– .			
			Spatacsin	
SDC12	10a12			appfirmed
36912	19413	AD	RTNZ	commed
SPG13	2q33.1	AD	HSPD1	confirmed
SPG14	3q27-q28	AR	Unknown	unconfirmed
SPG15	14g22-g24	AR	ZFYVE26	confirmed
SPG16	Xq11.2	XR	Unknown	unconfirmed
00047	11 - 12 2		DSCI 2	oo ofirmood
5PG17	11412.3	AD	DSULZ	continued
SPG18	8p11.23	AR	ERLIN2	confirmed
SPG19	9q33-q34	AD	Unknown	unconfirmed
SPG20	13a12.3	AR	Spartin	confirmed
SPG21	15q22.31	AR	ACP33	confirmed
00000	V-40.0		0101040	
5PG22	Xq13.2	XR	SLUTOAZ	confirmed
SPG23	1q24-q32	AR	Unknown	confirmed

SPG24	13q14	AR	Unknown	unconfirmed
SPG25	6q23.3- q24.1	AR	Unknown	unconfirmed
SPG26	12p11.1- q14	AR	B4GALNT1	confirmed
SPG27	10q22.1- q24.1	AR	Unknown	unconfirmed
SPG28	14q22.1	AR	DDHD1	confirmed
SPG29	1p31.1-21.1	AD	Unknown	unconfirmed
SPG30	2q37	AR	KIF1A	confirmed
SPG31	9p21	AD	REEP1	confirmed
SPG32	14q12-q21	AR	Unknown	unconfirmed
SPG33	10q24.2	AD	ZFYVE27	confirmed
SPG34	Xq25	XR	Unknown	unconfirmed
SPG35	16q23.1	AR	FA2H	confirmed
SPG36	12q23-24	AD	Unknown	unconfirmed
SPG37	8p21.1- q13.3	AD	Unknown	unconfirmed

SPG38	4p16-p15	AD	Unknown	unconfirmed
SPG39	19p13.2	AR	PNPLA6/NT E	confirmed
SPG40	Unknown, several loci excluded	AD	Unknown	unconfirmed
SPG41	11p14.1- p11.2	AD	Unknown	unconfirmed
SPG42	3q24-q26	AD	SLC33A1	unconfirmed
SPG43	19q12	AR	C19orf12	confirmed
SPG44	1q42.13	AR	GJC2 or GJA12	unconfirmed
SPG45	10q24.3- q25.1	AR	NT5C2	confirmed
SPG46	9p13.3	AR	GBA2	confirmed
SPG47	1p13.2-	AR	AP4B1	confirmed
SPG48	7p22.1	AR	KIAA0415	confirmed
SPG49	14q32.31	AR	TECPR2	confirmed

SPG50	7q22.1	AR	AP4M1	confirmed
SPG51	15q21.2	AR	AP4E1	confirmed
SPG52	14q12	AR	AP4S1	unconfirmed
SPG53	8p22	AR	Vps37A	unconfirmed
SPG54	8p11.23	AR	DDHD2 <sup>15</sup>	confirmed
SPG55	12q24	AR	C12orf65	confirmed
SPG56	4q25	AR	CYP2U1	unconfirmed
SPG57	3q12.2	AR	TFG <sup>16</sup>	unconfirmed
SPG58	17p13.2	AR	KIF1C <sup>17</sup>	confirmed
SPG59	15q21.2	AR	USP8 <sup>17</sup>	unconfirmed
SPG60	3p22.2	AR	WDR48 <sup>17</sup>	unconfirmed
SPG61	16p12.3	AR	ARL6IP1 <sup>17</sup>	unconfirmed
SPG62	10q24.31	AR	ERLIN1 <sup>17</sup>	confirmed
SPG63	1p13.3	AR	AMPD2 <sup>17</sup>	unconfirmed
SPG64	10q24.1	AR	ENTPD1 <sup>17</sup>	unconfirmed
SPG65	10q24.32	AR	NT5C2 <sup>17</sup>	Also designated SPG45

SPG66	5q32	AR	ARSI <sup>17</sup>	unconfirmed
SPG67	2q33.1	AR	PGAP1 <sup>17</sup>	unconfirmed
SPG68	11q13.1	AR	FLRT1 <sup>17</sup>	unconfirmed
SPG69	1q41	AR	RAB3GAP2	unconfirmed
SPG70	12q13.3	AR	MARS <sup>17</sup>	unconfirmed
SPG71	5p13.3	AR	ZFR <sup>17</sup>	unconfirmed
SPG72	5q31.2	AD/AR	REEP2 <sup>18</sup>	unconfirmed

AD, autosomal dominant; AR, autosomal recessive, XR, X chromosome recessive.

Supplementary Table 7: The previous list of locus symbols for primary familial brain calcification<sup>19, 20</sup>

Symbol	Gene locus	Inheritance	Gene symbol	Status and remarks
IBGC1	14q13	AD	-	Incorrect - Family later found to have a mutation in SLC20A2.
IBGC2	2q37	AD	-	Linkage to this locus found in one family. <sup>21</sup> Awaits independent confirmation.
IBGC3	8p21.1– 8q11.23	AD	SLC20A2	Confirmed
IBGC4	5q32	AD	PDGFRB	Mutations found in one family, segregating with phenotype and one sporadic case. <sup>22</sup> Awaits independent confirmation.
	22q13	AD	PDGFB	Mutations found in six families of varying ethnic origins. <sup>23</sup> Awaits independent confirmation. No locus symbol assigned.
	1q25.1	AD	XPR1	Mutations found in one family <sup>24</sup>

		Awaits confirmation. No locus
		symbol assigned

Supplementary Table 8: The current list of neurodegeneration with brain iron accumulation disorders<sup>25</sup>

Symbol	Gene locus	Inheritance	Gene	Status and remarks
			symbol	
NBIA1	20p13	AR	PANK2	Confirmed, also called
				PKAN
	22012		PLA2C6	Confirmed also called
INDIAZ	22412		FLA200	
				PARK14, PLAN
NBIA3	19q13	AD	FTL	Confirmed, also called
				neuroferritinopathy
NBIA4	19q12	AR	C19orf12	Confirmed, also called
				SPG43 and MPAN
NBIA5	Xp11.23	X-linked	WDR45	Confirmed also called
		dominant	(also	SENDA and BPAN.
			known as	
			WIPI4)	
Symbol	Gene locus	Inheritance	Gene	Status and remarks
			symbol	
NBIA1	20p13	AR	PANK2	Confirmed, also called

				PKAN
NBIA2	22q12	AR	PLA2G6	Confirmed, also called
				PARK14, PLAN
NBIA3	19q13	AD	FTL	Confirmed, also called
				neuroferritinopathy
	40-40		040-5-540	
NBIA4	19012	AR	C190/f12	Confirmed, also called
				SPG43 and MPAN
NBIA5	Xp11.23	X-linked	WDR45	Confirmed also called
		dominant	(also	SENDA and BPAN.
			known as	
			WIPI4)	

FA2H = fatty acid 2-hydroxylase, FTL = ferritin light chain, MPAN – mitochondrial membrane-associated neurodegeneration; NBIA = Neurodegeneration with brain iron accumulation, PANK2 = Pantothenate kinase 2, PKAN = pantothenate kinase-associated neurodegeneration, PLA2G6 = phospholipase A2, PLAN = PLA2G2-associated neurodegeneration, SENDA = static encephalopathy of childhood with neurodegeneration in adulthood, BPAN = Beta-propeller protein-associated neurodegeneration ; SPG = spastic paraplegia.

## References

1. Bonifati V. Genetics of Parkinson's disease--state of the art, 2013. Parkinsonism & Related Disorders 2014;20 Suppl 1:S23-28.

2. Trinh J, Farrer M. Advances in the genetics of Parkinson disease. Nat Rev Neurol 2013;9:445-454.

3. Klein C. Genetics in dystonia. Parkinsonism & Related Disorders 2014;20 Suppl 1:S137-142.

4. Zech M, Castrop F, Schormair B, et al. DYT16 revisited: exome sequencing identifies PRKRA mutations in a European dystonia family. Mov Disord 2014;29:1504-1510.

5. Jen JC. Hereditary episodic ataxias. Annals of the New York Academy of Sciences 2008;1142:250-253.

6. Cader MZ, Steckley JL, Dyment DA, McLachlan RS, Ebers GC. A genome-wide screen and linkage mapping for a large pedigree with episodic ataxia. Neurology 2005;65:156-158.

7. Kerber KA, Jen JC, Lee H, Nelson SF, Baloh RW. A new episodic ataxia syndrome with linkage to chromosome 19q13. Archives of Neurology 2007;64:749-752.

8. Verbeek DS, van de Warrenburg BP. The autosomal dominant cerebellar ataxias. Seminars in Neurology 2011;31:461-469.

9. Van Gaalen J, Giunti P, van de Warrenburg BP. Movement disorders in spinocerebellar ataxias. Mov Disord 2011;26:792-800.

10. Di Gregorio E, Borroni B, Giorgio E, et al. ELOVL5 mutations cause spinocerebellar ataxia 38. American Journal of Human Genetics 2014;95:209-217.

11. Tsoi H, Yu ACS, Chen ZS, et al. A novel missense mutation in CCDC88C activates the JNK pathway and causes a dominant form of spinocerebellar ataxia. Journal of Medical Genetics 2014.

12. Schneider SA, Walker RH, Bhatia KP. The Huntington's disease-like syndromes: what to consider in patients with a negative Huntington's disease gene test. Nat Clin Pract Neurol 2007;3:517-525.

13. Fink JK. Hereditary spastic paraplegia: clinico-pathologic features and emerging molecular mechanisms. Acta Neuropathol (Berl) 2013;126:307-328.

14. Salinas S, Proukakis C, Crosby A, Warner TT. Hereditary spastic paraplegia: clinical features and pathogenetic mechanisms. Lancet Neurology 2008;7:1127-1138.

15. Gonzalez M, Nampoothiri S, Kornblum C, et al. Mutations in phospholipase DDHD2 cause autosomal recessive hereditary spastic paraplegia (SPG54). Eur J Hum Genet 2013;21:1214-1218.

16. Beetz C, Johnson A, Schuh AL, et al. Inhibition of TFG function causes hereditary axon degeneration by impairing endoplasmic reticulum structure. Proc Natl Acad Sci U S A 2013;110:5091-5096.

17. Novarino G, Fenstermaker AG, Zaki MS, et al. Exome sequencing links corticospinal motor neuron disease to common neurodegenerative disorders. Science 2014;343:506-511.

18. Esteves T, Durr A, Mundwiller E, et al. Loss of association of REEP2 with membranes leads to hereditary spastic paraplegia. American Journal of Human Genetics 2014;94:268-277.

19. Hsu SC, Sears RL, Lemos RR, et al. Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics 2013;14:11-22.

20. Wang C, Li Y, Shi L, et al. Mutations in SLC20A2 link familial idiopathic basal ganglia calcification with phosphate homeostasis. Nature Genetics 2012;44:254-256.

21. Volpato CB, De Grandi A, Buffone E, et al. 2q37 as a susceptibility locus for idiopathic basal ganglia calcification (IBGC) in a large South Tyrolean family. Journal of Molecular Neuroscience 2009;39:346-353.

22. Nicolas G, Pottier C, Maltete D, et al. Mutation of the PDGFRB gene as a cause of idiopathic basal ganglia calcification. Neurology 2013;80:181-187.

23. Keller A, Westenberger A, Sobrido MJ, et al. Mutations in the gene encoding PDGF-B cause brain calcifications in humans and mice. Nature Genetics 2013;45:1077-1082.

Giovannini D, Legati A, Sitbon M, Geschwind DH, Coppola G, Battini J-L.
Identification of a mutation in the XPR1 gene associated with primary familial brain calcification. 7th SFB35 Symposium; 2014; Vienna: Medizinische Universitat Wien.
Doorn JM, Kruer MC. Newly characterized forms of neurodegeneration with brain iron accumulation. Current Neurology & Neuroscience Reports 2013;13:413.