Supplementary materials for: "The history of gene hunting in hereditary spinocerebellar degeneration: lessons from the past and future perspectives" by Yahia & Stevanin.

Supplementary Table 1. Methods used in identifying the subtypes of hereditary spinocerebellar disorders, excluding syndromic disorders in which ataxia and/or spastic paraplegia are not prominent clinical features and unconfirmed clinico-genetic entities (SPG59, 60, 65, 67, 68, 69, 70 and 71).

SPG: spastic paraplegia; LEMSPAD: leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome; SCA: spinocerebellar ataxia; EA: episodic ataxia; SCA42ND: spinocerebellar ataxia type 42, early-onset, severe, with neurodevelopmental deficits; SPAX: spastic ataxia; DRPLA: dentatorubral-pallidoluysian atrophy; ADCADN: cerebellar ataxia, deafness, and narcolepsy, autosomal dominant; CAPOS: cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss; BNHS: Boucher-Neuhauser syndrome; SPOAN: spastic paraplegia, optic atrophy, and neuropathy; NEDSWMA: neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities; SPPRS: Spastic paraplegia and psychomotor retardation with or without seizures; HHH syndrome: Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome; EAOH: ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia; AT: ataxia-telangiectasia; FRDA: Friedreich ataxia; CANVAS: cerebellar ataxia, neuropathy, and vestibular areflexia syndrome; AOA: ataxia-oculomotor apraxia; MSS: Marinesco-Sjogren syndrome; VED: Ataxia with isolated vitamin E deficiency; PHARC: polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract; ICRD: infantile cerebellar-retinal degeneration; ATCAY: ataxia, cerebellar, Cayman type; CAMRQ: cerebellar ataxia, mental retardation, and dysequilibrium syndrome; LKPAT: leukoencephalopathy with ataxia; SESAMES: seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance syndrome; PTBHS: Poretti-Boltshauser syndrome; PACA: pancreatic and cerebellar agenesis; GDHS: Gordon Holmes syndrome; NADGP: neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset; MTDPS7: Mitochondrial DNA depletion syndrome 7; SCAE: Spinocerebellar ataxia with epilepsy; SCAR: autosomal recessive spinocerebellar ataxia; SCAN: autosomal recessive spinocerebellar ataxia with axonal neuropathy; ISQMR: ichthyosis, spastic quadriplegia, and mental retardation; ASAT: sideroblastic anemia with ataxia; SCAX1: spinocerebellar ataxia, X-linked 1; FXTAS: fragile X tremor/ataxia syndrome; LA: linkage analysis; CA: candidate gene approach; WES: whole-exome sequencing; TE: targeted next generation sequencing; WGS: whole-genome sequencing; Repeat-exp: repeat expansion detection approaches. #: molecular cause known, %: molecular cause unknown.

Disease	Gene	OMIM	Method	Reference
Autosomal dominant/de novo				
SPG4	*SPAST*	# 182601	LA, CA	(Hazan et al. 1999)
SPG6	*NIPA1*	# 600363	LA, CA	(Rainier et al. 2003)
SPG8	*WASHC5*	# 603563	LA, CA	(Valdmanis et al. 2007)
SPG10	*KIF5A*	# 604187	LA, CA	(Reid et al. 2002)
SPG12	*RTN2*	# 604805	WES	(Montenegro et al. 2012)
SPG13	*HSPD1*	# 605280	LA, CA	(Hansen et al. 2002)
SPG17	*BSCL2*	# 270685	LA, CA	(Windpassinger et al. 2004)
SPG19		% 607152	LA	(Orlacchio et al. 2005)
SPG29		% 609727	LA	(Orlacchio et al. 2005)
SPG31	*REEP1*	# 610250	LA, CA	(Züchner et al. 2006)

SPG36		**%** 613096	LA	(Schüle et al. 2009)
SPG38		**%** 612335	LA	(Orlacchio et al. 2008)
SPG41		**%** 613364	LA	(Zhao et al. 2008)
SPG42	*SLC33A1*	# 612539	LA, CA	(Lin et al. 2008)
SPG73	*CPT1C*	# 616282	LA, WES	(Rinaldi et al. 2015)
SPG80	*UBAP1*	# 618418	WES	(Farazi Fard et al. 2019)
LEMSPAD	*EIF2AK1*	# 618878	WES	(Mao et al. 2020)
TUBB4A-SPG	*TUBB4A*		TE	(Sagnelli et al. 2016)
BICD2-SPG	*BICD2*		LA, WES, DNA array	(Oates et al. 2013)
SCA1	*ATXN1*	# 164400	LA, Repeat-exp	(Banfi et al. 1994)
SCA2	*ATXN2*	# 183090	LA, Repeat-exp	(Pulst et al. 1996)
SCA3	*ATXN3*	# 109150	LA, Repeat-exp	(Kawaguchi et al. 1994)
SCA4		**%** 600223	LA	(Hellenbroich et al. 2003)
SCA6	*CACNA1A*	# 183086	CA, Repeat-exp	(Zhuchenko et al. 1997)
EA2		# 108500	LA, CA	(Ophoff et al. 1996)
SCA7	*ATXN7*	# 164500	LA, Repeat-exp	(David et al. 1997)
SCA8	*ATXN8OS*	# 608768	Repeat-exp	(Koob et al. 1999)
	ATXN8		CA	(Moseley et al. 2006)
SCA9		**%** 612876		(Higgins et al. 1997)
SCA10	*ATXN10*	# 603516	LA, Repeat-exp	(Matsuura et al. 2000)
SCA11	*TTBK2*	# 604432	LA, CA	(Houlden et al. 2007)
SCA12	*PPP2R2B*	# 604326	LA, Repeat-exp	(Holmes et al. 1999)
SCA13	*KCNC3*	# 605259	LA, CA	(Waters et al. 2006)
SCA14	*PRKCG*	# 605361	LA, CA	(Chen et al. 2003)
SCA15	*ITPR1*	# 606658	LA, CA, DNA array	(Van De Leemput et al. 2007)
SCA29		# 117360	LA, WES	(Huang et al. 2012)
SCA17	*TBP*	# 607136	CA, Repeat-exp	(Reiji Koide et al. 1999)
SCA18		**%** 607458	LA	(Brkanac et al. 2002)
SCA19	*KCND3*	# 607346	LA, WES	(Lee et al. 2012)
SCA20		# 608687	LA, DNA array	(Knight et al. 2008)
SCA21	*TMEM240*	# 607454	LA, WES	(Delplanque et al. 2014)
SCA23	*PDYN*	# 610245	LA, CA	(Bakalkin et al. 2010)
SCA25		**%** 608703	LA	(Stevanin et al. 2004)
SCA26	*EEF2*	# 609306	LA, TE	(Hekman et al. 2012)
SCA27	*FGF14*	# 609307	LA, CA	(Van Swieten et al. 2003)
SCA30		**%** 613371	LA	(Storey et al. 2009)
SCA31	*BEAN1*	# 117210	LA, CA	(Sato et al. 2009)
SCA35	*TGM6*	# 613908	LA, WES, DNA array	(Wang et al. 2010)
SCA36	*NOP56*	# 614153	LA, CA	(Kobayashi et al. 2011)
SCA37	*DAB1*	# 615945	LA, TE, DNA array	(Seixas et al. 2017)
SCA38	*ELOVL5*	# 615957	LA, TE, DNA array	(Di Gregorio et al. 2014)
SCA40	*CCDC88C*	# 616053	WES	(Tsoi et al. 2014)
SCA41	*TRPC3*	# 616410	WES	(Fogel et al. 2015)

SCA42	*CACNA1G*	# 616795	LA, WES, DNA array	(Coutelier, Blesneac, et al. 2015)
SCA42ND		# 618087	WES	(Chemin et al. 2018)
SCA43	*MME*	# 617018	LA, WES, DNA array	(Depondt et al. 2016)
SCA45	*FAT2*	# 617769	WES	(Nibbeling et al. 2017)
SCA46	*PLD3*	# 617770	WES	(Nibbeling et al. 2017)
SCA47	*PUM1*	# 617931	CA, WES, DNA array	(Gennarino et al. 2018)
SPAX1	*VAMP1*	# 108600	LA, CA	(Bourassa et al. 2012)
KCNA2-SPG and ataxia	*KCNA2*		WES	(Helbig et al. 2016)
DRPLA	*ATN1*	# 125370	Repeat-exp	(R. Koide et al. 1994)
ADCADN	*DNMT1*	# 604121	WES	(Winkelmann et al. 2012)
CAPOS	*ATP1A3*	# 601338	WES	(Demos et al. 2014)
EA1	*KCNA1*	# 160120	LA, CA	(Browne et al. 1994)
EA3		% 606554	LA	(Steckley et al. 2001)
EA5	*CACNB4*	# 613855	CA	(Escayg et al. 2000)
EA6	*SLC1A3*	# 612656	CA	(Jen et al. 2005)
EA7		% 611907	LA	(Kerber et al. 2007)
EA8		% 616055	LA	(Conroy et al. 2014)
EA9	*SCN2A*	# 618924	CA	(Liao et al. 2010)
Autosomal Recessive				
SPG5A	*CYP7B1*	# 270800	LA, CA	(Tsaousidou et al. 2008)
SPG11	*SPG11*	# 604360	LA, CA	(Stevanin et al. 2007)
SPG14		% 605229	LA	(Vazza et al. 2000)
SPG15	*ZFYVE26*	# 270700	LA, CA	(Hanein et al. 2008)
SPG20	*SPG20*	# 275900	LA, CA	(Patel et al. 2002)
SPG21	*ACP33*	# 248900	LA, CA	(Simpson et al. 2003)
SPG23	*DSTYK*	# 270750	WES	(J. Y. W. Lee et al. 2017)
SPG24		% 607584	LA	(Hodgkinson et al. 2002)
SPG25		% 608220	LA	(Zortea et al. 2002)
SPG26	*B4GALNT1*	# 609195	LA, WES	(Boukhris et al. 2013)
SPG27		% 609041	LA	(Meijer et al. 2004)
SPG28	*DDHD1*	# 609340	LA, TE	(Tesson et al. 2012)
SPG32		% 611252	LA	(Stevanin et al. 2007)
SPG35	*FA2H*	# 612319	DNA array, CA	(Edvardson et al. 2008)
SPG39	*PNPLA6*	# 612020	LA, CA	(Rainier et al. 2008)
BNHS		# 215470	WES	(Synofzik et al. 2014)
SPG43	*C19orf12*	# 615043	WES	(Landouré et al. 2013)
SPG44	*GJC2*	# 613206	CA	(Orthmann-Murphy et al. 2009)
SPG45	*NT5C2*	# 613162	WES	(Novarino et al. 2014)
SPG46	*GBA2*	# 614409	WES	(Martin et al. 2013)
SPG47	*AP4B1*	# 614066	LA, CA	(Abou Jamra et al. 2011)
SPG48	*AP5Z1*	# 613647	CA	(Słabicki et al. 2010)

SPG49	*TECPR2*	# 615031	WES	(Oz-Levi et al. 2012)
SPG50	*AP4M1*	# 612936	LA, CA	(Verkerk et al. 2009)
SPG51	*AP4E1*	# 613744	DNA array	(Moreno-De-Luca et al. 2011)
SPG52	*AP4S1*	# 614067	LA, WES	(Abou Jamra et al. 2011)
SPG53	*VPS37A*	# 614898	LA, CA	(Zivony-Elboum et al. 2012)
SPG54	*DDHD2*	# 615033	WES	(Schuurs-Hoeijmakers et al. 2012)
SPG55	*C12orf65*	# 615035	LA, WES, DNA array	(Shimazaki et al. 2012)
SPG56	*CYP2U1*	# 615030	LA, CA, DNA array	(Tesson et al. 2012)
SPG57	*TFG*	# 615658	LA, WES, DNA array	(Beetz et al. 2013)
SPG61	*ARL6IP1*	# 615685	WES	(Novarino et al. 2014)
SPG62	*ERLIN1*	# 615681	WES	(Novarino et al. 2014)
SPG63	*AMPD2*	# 615686	WES	(Novarino et al. 2014)
SPG64	*ENTPD1*	# 615683	WES	(Novarino et al. 2014)
SPOAN	*KLC2*	# 609541	WGS	(Melo et al. 2015)
SPG74	*IBA57*	# 616451	LA, WES	(Lossos et al. 2015)
SPG75	*MAG*	# 616680	WES	(Novarino et al. 2014)
SPG76	*CAPN1*	# 616907	WES	(Gan-Or et al. 2016)
SPG77	*FARS2*	# 617046	WES, DNA array	(Yang et al. 2016)
SPG78	*ATP13A2*	# 617225	WES	(Kara et al. 2016)
SPG79	*UCHL1*	# 615491	WES, DNA array	(Bilguvar et al. 2013)
SPG81	*SELENOI*	# 618768	LA, DNA array, WES	(Ahmed et al. 2017)
SPG82	*PCYT2*	# 618770	WES	(Vaz et al. 2019)
SPG83	*HPDL*	# 619027	WES	(Husain et al. 2020)
NEDSWMA		# 619026	WES	(Husain et al. 2020)
Hereditary sensory neuropathy with spastic paraplegia	*CCT5*	# 256840	LA, CA	(Bouhouche et al. 2006)
CTX	*CYP27A1*	# 213700	CA	(Cali et al. 1991)
SPPRS	*HACE1*	# 616756	DNA array, WES	(Hollstein et al. 2015)
HHH syndrome	*SLC25A15*	# 238970	CA	(Camacho et al. 1999)
Argininemia	*ARG1*	# 207800	CA	(Haraguchi et al. 1990)
DARS2-SPG	*DARS2*		CA	(Lan et al. 2017)
PLA2G6-SPG	*PLA2G6*		WES, TE	(Ozes et al. 2017)
GPT2-SPG	*GPT2*		WES	(Kaymakcalan et al. 2018)
ADCY5-SPG	*ADCY5*		WES, TE	(Waalkens et al. 2018)
RNF170-SPG	*RNF170*		WES, WGS	(Wagner et al. 2019)
LYST-SPG	*LYST*		LA, WES, DNA array	(Shimazaki et al. 2014)

EAOH	*APTX*	# 208920	LA, CA	(Date et al. 2001)
AT	*ATM*	# 208900	LA, CA	(Savitsky et al. 1995)
AT-like disorder 1	*MRE11*	# 604391	CA	(Stewart et al. 1999)
Nijmegen breakage syndrome	*NBN*	# 251260	LA, CA	(Varon et al. 1998)
Cockayne syndrome, type A	*ERCC8*	# 216400	CA	(Henning et al. 1995)
Cockayne syndrome, type B	*ERCC6*	# 133540	CA	(Troelstra et al. 1992)
Leukoencephalopathy with vanishing white matter	*EIF2B1*	# 603896	CA	(Van Der Knaap et al. 2002)
	EIF2B2		LA, CA	(Leegwater et al. 2001)
	EIF2B3		CA	(Van Der Knaap et al. 2002)
	EIF2B4		CA	(Van Der Knaap et al. 2002)
	EIF2B5		LA, CA	(Leegwater et al. 2001)
FRDA	*FXN*	# 229300	LA, CA	(Campuzano et al. 1996)
HARS1-ataxia	*HARS1*		WES, TE	(Galatolo et al. 2020)
CANVAS	*RFC1*	# 614575	LA, DNA array, WGS	(Cortese et al. 2019)
SPAX2	*KIF1C*	# 611302	LA, DNA array, WES	(Dor et al. 2014)
SPAX3	*MARS2*	# 611390	LA, DNA array, CA	(Bayat et al. 2012)
SPAX6	*SACS*	# 270550	LA, CA	(Engert et al. 2000)
SPAX8	*NKX6-2*	# 617560	WES	(Chelban et al. 2017)
SPAX9	*CHP1*	# 618438	LA, DNA array, WES	(Mendoza-Ferreira et al. 2018)
POLR3A-SPAX	*POLR3A*		LA, DNA array, WES	(Minnerop et al. 2017)
AOA3	*PIK3R5*	# 615217	LA, DNA array, CA	(Tassan et al. 2012)
AOA4	*PNKP*	# 616267	DNA array, CA	(Bras et al. 2015)
MSS	*SIL1*	# 248800	LA, CA	(Anttonen et al. 2005)
VED	*TTPA*	# 277460	LA, CA	(Ouahchi et al. 1995)
PHARC	*ABHD12*	# 612674	LA, CA	(Fiskerstrand et al. 2010)
ICRD	*ACO2*	# 614559	LA, DNA array, WES	(Spiegel et al. 2012)
ATCAY	*ATCAY*	# 601238	LA, CA	(Bomar et al. 2003)
CAMRQ1	*VLDLR*	# 224050	LA, CA	(Boycott et al. 2005)
CAMRQ4	*ATP8A2*	# 615268	DNA array, TE	(Onat et al. 2013)
LKPAT	*CLCN2*	# 615651	WES	(Depienne et al. 2013)
SESAMES	*KCNJ10*	# 612780	LA, CA	(Scholl et al. 2009)
PTBHS	*LAMA1*	# 615960	DNA array, WES	(Aldinger et al. 2014)
PACA	*PTF1A*	# 609069	LA, CA	(Sellick et al. 2004)

GDHS	*RNF216*	# 212840	WES, TE	(Margolin et al. 2013)
NADGP	*SQSTM1*	# 617145	WES	(Haack et al. 2016)
PMM2-ataxia	*PMM2*		WES	(Van De Warrenburg et al. 2016)
SCAE	*POLG*	# 607459	CA	(Winterthun et al. 2005)
SCAR2	*PMPCA*	# 213200	LA, CA, WES	(Jobling et al. 2015)
SCAR3		**%** 271250	LA	(Bomont et al. 2000)
SCAR4	*VPS13D*	# 607317	LA, DNA array, WES	(Seong et al. 2018)
SCAR6		**%** 608029	LA	(Tranebjaerg et al. 2003)
SCAR7	*TPP1*	# 609270	LA, WES	(Sun et al. 2013)
SCAR8	*SYNE1*	# 610743	LA, CA	(Gros-Louis et al. 2007)
SCAR9	*ADCK3*	# 612016	CA	(Mollet et al. 2008)
SCAR10	*ANO10*	# 613728	LA, DNA array, TE	(Vermeer et al. 2010)
SCAR11	*SYT14*	# 614229	WES	(Doi et al. 2011)
SCAR12	*WWOX*	# 614322	WES	(Mallaret et al. 2014)
SCAR15	*RUBCN*	# 615705	LA, CA	(Assoum et al. 2010)
SCAR17	*CWF19L1*	# 616127	LA, DNA array, WES	(Burns et al. 2014)
SCAR19	*SLC9A1*	# 616291	DNA array, WES	(Guissart et al. 2015)
SCAR20	*SNX14*	# 616354	DNA array, WES	(Thomas et al. 2014)
SCAR21	*SCYL1*	# 616719	WES	(Schmidt et al. 2015)
SCAR22	*VWA3B*	# 616948	DNA array, WES	(Kawarai et al. 2016)
SCAR23	*TDP2*	# 616949	DNA array, WES	(Gómez-Herreros et al. 2014)
SCAR24	*UBA5*	# 617133	WES	(Duan et al. 2016)
SCAR25	*ATG5*	# 617584	LA, DNA array, WES	(Kim et al. 2016)
SCAR26	*XRCC1*	# 617633	WES	(Hoch et al. 2017)
SCAR27	*GDAP2*	# 618369	WES	(Eidhof et al. 2018)
SCAR28	*THG1L*	# 618800	WES	(Edvardson et al. 2016)
SCAN1	*TDP1*	# 607250	LA, CA	(Takashima et al. 2002)
SCAN2	*SETX*	# 606002	LA, CA	(Moreira et al. 2004)
SCAN3	*COA7*	# 618387	WES	(Lyons et al. 2016)
Dual autosomal dominant and recessive transmissions				
Dominant SPG3A	*ATL1*	# 182600	LA, CA	(Zhao et al. 2001)
ATL1-rcessive SPG			WES	(Khan et al. 2014)
Dominant SPG7	*PGN*	# 607259	CA	(Sánchez-Ferrero et al. 2013)
Recessive SPG7			LA, CA	(Casari et al. 1998)
SPG9A (dominant)	*ALDH18A1*	# 601162	LA, WES, TE, DNA array	(Coutelier et al. 2015)
SPG9B		# 616586		

(recessive)				
Recessive SPG18	*ERLIN2*	# 611225	LA, CA, DNA array	(Yildirim et al. 2011)
ERLIN2-dominant SPG			WES	(Rydning et al. 2018)
SPG30 (dominant)	*KIF1A*	# 610357	TE	(Ylikallio et al. 2015)
SPG30 (recessive)			WES, DNA array	(Erlich et al. 2011)
SPG72 (dominant)	*REEP2*	# 615625	LA, WES, DNA array	(Esteves et al. 2014)
REEP2-recessive SPG				
SCA5 (dominant)	*SPTBN2*	# 600224	LA, Repeat-exp	(Ikeda et al. 2006)
SCAR14 (recessive)		# 615386	TE	(Lise et al. 2012)
SCA28 (dominant)	*AFG3L2*	# 610246	LA, CA	(Di Bella et al. 2010)
SPAX5 (recessive)		# 614487	WES	(Pierson et al. 2011)
SCA34 (dominant)	*ELOVL4*	# 133190	LA, WES	(Cadieux-Dion et al. 2014)
ISQMR (recessive)		# 614457	WES, DNA array	(Aldahmesh et al. 2011)
SCA44 (dominant)	*GRM1*	# 617691	WES	(Watson et al. 2017)
SCAR13 (recessive)		# 614831	LA, WES	(Guergueltcheva et al. 2012)
SCA48 (dominant)	*STUB1*	# 618093	LA, WES	(Genis et al. 2018)
SCAR16 (recessive)		# 615768	LA, WES, DNA array	(Shi et al. 2013)
GRID2-ataxia (dominant)	*GRID2*		LA, TE, WES, DNA array	(Coutelier et al. 2015)
SCAR18 (recessive)		# 616204	DNA array	(Utine et al. 2013)
X-linked				
SPG1	*L1CAM*	# 303350	LA, CA	(Jouet et al. 1994)
SPG2	*PLP1*	# 312920	LA, CA	(Saugier-Veber et al. 1994)
SPG16		**%** 300266	LA	(Steinmuller et al. 1997)
SPG22	*SLC16A2*	# 300523	CA	(Friesema et al. 2004)
SPG34		**%** 300750	LA	(Macedo-Souza et al. 2008)
ASAT	*ABCB7*	# 301310	LA, CA	(Allikmets et al. 1999)
SCAX1	*ATP2B3*	# 302500	TE	(Zanni et al. 2012)
FXTAS	*FMR1*	# 300623	CA	(Hagerman et al. 2001)

Mitochondrial				
SPAX4	*MTPAP*	# 613672	LA, DNA array, CA	(Crosby et al. 2010)
Mitochondrial complex V deficiency	*MTATP6*		CA	(Verny et al. 2011)
MTDPS7	*TWNK*	# 271245	CA	(Sarzi et al. 2007)

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