

**Supplementary table 1: Genotype-phenotype correlation of participants**

	Sex	AT subtype	Mutation(s)	ATM protein	ATM kinase	AFP (kU/L)	Phenotype	SARA (/40)	Immunoglobulins	Cancer	WMH (Schelten's score/Fazekas)	CM Bs
1	M	Classic	c.3576G>A;p.Ser1135_Lys1192del58 (skip of exon 26) c.2413C>T;p.Arg805Ter.	Absent	n.d	297	Ataxia, Dystonic grimacing, myoclonic jerks, dystonic upper limb posturing, severe neuropathy	34	Normal profile	N	0/0	0
2	M	Classic	c.6047A>G p.Asp2016Gly c.3576 G>A p.Ser1135_Lys1192del58	Low	None	124	Ataxia, Facial grimacing, Chorea, dystonic tongue movements, Myoclonic jerks, balismus. Moderate – severe neuropathy	36	Normal profile	N	0/0	0
3	M	Classic	c.1158_delG;p.Arg387fs c.5623C>T;p.Arg1875*.	Absent	None	116	Ataxia. No significant dystonia. Occasional myoclonic jerks. Moderate neuropathy	35	IgA deficiency, IgM increase. Normal IgG subclasses	N	8/1	153
4	F	Classic	c.1442delT;p.Leu481* c.9022C>T;p.Arg3008Cys	50% protein	None	432	Ataxia, upper and lower limb dystonia, tongue. Moderate – severe neuropathy	33	Normal profile	Follicular hemithyroidectomy	4/1	n.d
5	F	Classic	c.7013T>C p.Leu2338Pro c.6056A>G p.Tyr2019Cys	Low protein	None	165	Ataxia, upper and lower limb dystonia, facial grimacing. Moderate neuropathy	29	Elevated IgM, IgA deficiency	N	0/0	0
6	F	Classic	Homozygous c.1262C>A;p.Ser421Ter	Absent	None	359	Mild ataxia, Significant dystonic and myoclonic jerks, dystonic posturing hands. Moderate – severe	30	Low IgA, mild polyclonal increase IgM, IgG2 deficiency	N	0/0	1

							neuropathy					
7	M	Classic	c.5290_5290delC; p.Leu1764fs c.2250 G>A; p.Glu709_Lys750del42(loss of exon 16)	Trace	None	460	Ataxia. Face upper and lower limb dystonia & myoclonus, moderate neuropathy	26	Hypogammaglobulinemia	N	3/0	110
8	F	Classic	c.7913G>A, p.Trp2638*exon55 c.5848G>C, p.A1a1950 pro	Low level ATM protein about 10%	None	491	Ataxia. Myoclonic jerks, dystonia. Hyperkinetic movements head, trunk, limbs. Slightly slow tongue movements, severe neuropathy	26	Polyclonal IgM, reduced IgG2, N IgA	N	2/1	1
9	M	Classic	c.5363delG p.Gly1788fs c.2251-2A>T p.Ser751_Lys792del loss of exon 17	Absent	n.d	620	Ataxia. Head trunk dystonia, upper limb myoclonus. Moderate neuropathy	37	IgG2, IgG3 subclass deficiency, mild pancytopenia	N	n.d - extensive oedema	121
10	F	Classic	c.5290delC;p.Leu1764fs c.1564_1565delGA,P.Glu522IlefsER43 exon 12	Absent	n.d	413	Ataxia. Face, neck, upper and lower limb dystonia, mild myoclonus, chorea. Moderate neuropathy	28	IgA deficiency, IgM normal	N	0/0	3
11	M	Classic	c.5623C>T;p.Arg1875 c.8305insA;p.Trp2769	Absent	None	446	Marked ataxia. Limb, truncal dystonia and significant jerks upper limbs. Moderate neuropathy	33	Hypogammaglobinemia, IgA, IgM deficiency	N	n.d	n.d
12	M	Classic	c.5515C>T p.Gln1839* IVS16-1G>C del19bp exon 17	Absent	n.d	390	Ataxia. Dystonic limbs. Neuropathy	29	Reduced IgA and IgG2, IgM normal	Dermatofibrosarcoma protuberans	5/0	10
13	F	Classic	c.5623C>T; p.Arg1875Ter c.2250 ins3; p.Lys750_Ser751ins3	Absent	None	268	Moderate truncal and limb ataxia. Severe neuropathy	27	Normal profile	N	n.d	n.d

14	F	Classic	c.4389_4389delT; p.Phe1463fs c.7928del83(del exon56).	Absent	None	312	Ataxia. Dystonia limbs, grimacing , occasional myoclonic jerks. Moderate – severe neuropathy	35	Undetectable IgA, hypogammaglobulinemia	N	1/0	0
15	M	Classic	c.2483delA p.Lys828SerfsTer8 c.3802delG p.Val1268Ter	Absent	None	373	Ataxia. Dystonic tremor upper limbs and posturing . Severe neuropathy	36	IgA deficiency, elevated IgM	N	2/0	1
16	M	Variant	c.5763-1050A>G;p.Pro1922fs c.8491del7	Very low level protein (5%)	Low	82	Ataxia. Dystonic posturing , myoclonic jerks. Moderate neuropathy	18	Normal profile	N	0/0	0
17	M	Variant	c.7665delinsGTGA;p.His2555GlnfsX2 c.2T>C;p.Met1Thr	Low	n.d	296	Ataxia. Upper and lower limb dystonia and chorea. Slow tongue movements, jerking neck. Moderate – severe neuropathy	30	IgA MGUS, IgM normal	N	n.d	n.d
18	F	Variant	c.6490G>T; p.Glu2164Ter c.7184A>T; p.Asp2395Val.	Very low	Some retained	26	Ataxia. Dystonic tremor. Severe neuropathy	23	Low IgA, low IgM and IgG4	N	3/1	n.d
19	F	Variant	c.1564_1565delGA;p.Glu522IlefsTer43(exon12) c.8480T>G;p.Phe2827Cys	Low (10%)	Some retained	386	Ataxia. Dystonia, moderate neuropathy	15	Polyclonal raised IgM, normal IgA	N	1/0	n.d
20	F	Variant	c.5763-1050A>G; p.Pro1922fs c.9022C>T; p.Arg3008Cys.	Present	Some retained	156	Ataxia. Cervical dystonia with persistent head truncal tremor, myoclonic jerks. Severe neuropathy	?	Slightly reduced IgG3	N	2/0	0
21	F	Variant	c.6490G>T;p.Glu2164Ter c.7184A>T; p.Asp2395Val	Low	Some retained	74	Ataxia. Moderate neuropathy	13	IgA deficiency, mild polyclonal increase IgM	N	1/0	0
22	F	Variant	c.8266A>T;p.Lys2756Ter c.7271T>G; p.vAl2424Gly	Present	Some	22	Ataxia. Dystonic posturing and tremor.	29	Normal profile	Neuroendocrine tumour	4/1	0

							Mild-moderate sensory neuropathy					
23	F	Variation	c.2T>C;p.Met1Thr c.5763-1050A>G;p.(Pro1992fs)	Low	Low	49	Ataxia. Dystonic head tremor. Severe neuropathy	33	Normal profile	N	3/1	0
24	F	Variation	c.1158delG;p.(Lys387fs) c.6807G7A(loss of exon48)	Low	Low A	126	Ataxia. Dystonia. Moderate neuropathy	23	Normal profile	Breast cancer -	0/0	n.d
25	M	Variation	c.9023G>A;p.Arg3008His c.1066-6T>G(loss of exon 11)	Near normal	Reduced	43	Ataxia. Myoclonus. No neuropathy	15	Normal profile	N	1/1	0
26	F	Variation	IVS18+6T>A Second mutation not identified.	Low	Some retained	2	Ataxia. Mild facial dyskinesia. Moderate – severe neuropathy	23	Normal profile	N	0/0	0
27	F	Variation	c.T2C;p.Met1Thr c.640_640delT;p.Ser214ProfsTer16	Low	None	291	Ataxia. Occasional myoclonic jerks, no significant extrapyramidal movements. Severe neuropathy	34	IgA and IgG2 subclass deficiency. Low normal IgM	N	3/0	n.d
28	F	Variation	c.5763-1050a>g;p.Pro1922fs c.6136delC;p.Leu2046fs*1.	Some retained	n.d	110	Ataxia. Mild dystonic posturing neck. Severe neuropathy	25	Mild polyclonal raised IgA, normal IgM	Dermatofibrosarcoma protuberans	4/0	0
29	M	Variation	c.8494C>T; p.Arg2832Cys c.1844T>C;p.Leu615Pro	Low	Low	715	Ataxia. Occasional myoclonic jerks, no significant extrapyramidal movements	25	Normal profile	N	1/0	3
30	F	Variation	c.5763-1050A>G. p.Pro1922fs c.1563_1564delA>G;p.Glu522Ilefs*21	Low	Some	54	Ataxia. Dystonic tremor neck and trunk, posturing upper limbs, occasional myoclonic jerks. Moderate neuropathy	20	Normal profile	No	1/0	0

31	M	Variant	c.170G>A;p.Trp57* c.3403del174 del exon26	Low	Some	89	Ataxia. Upper and lower limb dystonia. Grimacing. Severe neuropathy	35	Low IgM, IgA normal	N	4/0	0
32	M	Variant	c.170G>A;p.Trp57* c.3403del174 del exon26	Low	Some	56	Ataxia. Neck, upper and lower limb dystonia, upper limb weakness. Severe neuropathy	37	Normal profile	N	4/1	n.d.
33	F	Variant	homozygous c.590G>A;p.Gly197Gln	Very low	Reduced	419 -	No ataxia. Orofacial dystonia. No neuropathy	5	Slightly elevated IgG4	Breast cancer	4/1	0
34	M	Variant	c.1564_1565delGA;p.Glu522IlefsTer*43 c.8418+681A>G; p.Glu2807Valfs*4	Very low	Some	50	Ataxia. Mild upper and lower limb dystonia, facial grimacing, mild myoclonus. Mild-moderate neuropathy	19	Normal profile	N	2/1	0
35	F	Variant	c.5763-1050A>G;p.Pro1922Leufs*9 c.8418+2_5delTTGAG;p.Val2757_Met2806del	Low (10%)	Retained	332	Ataxia. Neck, upper and lower limb dystonia, myoclonus. Severe neuropathy	34	IgG3 deficiency, normal IgA, IgM	N	3/1	0
36	F	Variant	c.6490G>T;p.Glu2164Ter c.7184A>T; p.Asp2395Val	Low	Some retained	55	Ataxia. Dystonia and tremor	17	Low IgA, elevated IgM	Chronic myeloid leukaemia	11/1	0
37	M	Variant	c.3G>T;p.Met1Ile c.7271T>G;p.Val2424Gly	Reduced (50%)	Some retained	22	Ataxia. Myoclonus, orofacial dyskinesia. Moderate neuropathy	26.5	Mild elevated IgM, normal IgA	N	1/0	0
38	F	Variant	c.2730_2731insAG; p.Thr910fsTer c.5005_5006ins255; p.Gly1669GlyfsTer2	Low	Residual	3	Ataxia. Prominent dyskinesia in neck and upper limbs. Mild neuropathy	3	IgA deficiency, IgG2 normal, normal IgM	T cell acute lymphocytic leukaemia	0/0	0

**Supplementary table 1:** Genotype-phenotype correlation. Key: M = male, F = female, n.d = not done, N = no. MRI excluded if extensive motion artefact or no suitable sequence for analysis