

Supplementary data

Table 1: Clinical and Genetic data of patients

Patient	MT-2A	MT-1A	MT-1E	TBARS	CAT	Age (years)	Clinical phenotype	Protein mutation	Gene mutation	Group of mutation
1	2,72	65,45	5,87	0,06	143,52	9	Co	p-Val331GlyfsX34	c.1052_1188del137	LT
2	11,75	9,68	1,45	0,11	157,41	9	C	p.Arg168X	c 502 C>T	ET
3	2,54	69,61	0,52	0,13	226,39	5	Co	p.Arg270Glufs288X	c.808delC	IT
4	0,80	14,46	0,86			11	C	3-4 Exon deletion		ET
5	1,32	26,22	3,00	0,17	219,32	6	C	p.Arg255X	c.763C>T	IT
6	0,93	0,35	4,92	0,15	298,92	3	C	p.Lys135Asn	c.405A>C	M
7	1,06	19,69	1,79	0,11	182,26	14	C	p.Arg294X	c.880C>T	LT
8	1,43	54,76	2,55	0,21	226,09	3	C	p.Arg168X	c 502 C>T	ET
9	0,51	44,69	0,53	0,18	146,44	11	C	p.Leu386His fs390X	C.1156_1197del41	LT
10	1,39	1,16	6,29	0,13	271,26	6	C	p.Leu386His fs390X	C.1156_1197del41	LT
11	0,95	62,63	0,72	0,09	334,08	10	PSV	p.P387fsX9	c.1160_1166del7	LT
12	2,70	3,43	2,20	0,15	213,49	12	C	p.Arg306Cys	c 916 C>T	M
13	3,20	0,00	2,00	0,16	273,67	7	C	p.P388fsX	c.1163del26	LT
14	0,88	64,88	0,45	0,20	260,78	7	C	p.Lys144Arg fs2X	c.431delA	ET
15	6,00	4,65	8,04	0,20	256,19	6	C	p.Asp156Glu	c.468 C>G	M
16	2,87	60,06	4,96	0,17	252,34	4	C	p.Arg306Cys	c 916 C>T	M
17	4,51	50,04	6,91			4	C	p.Pro225Arg	c 674 C>G	M
18	1,98	46,09	3,66			4	C	p.Arg255X	c.763C>T	IT
19	1,25	0,58	2,42	0,08	190,78	4	C	p.Arg255X	c.763C>T	IT
20	0,45	9,59	1,28	0,20	301,89	3	C	p.Glu37Xfs	c.108-111 del	ET
21	3,96	52,37	2,88	0,14	238,25	6	C	p. Arg168X	c 502 C>T	ET
22	0,71	0,16	0,09	0,15	251,42	7	C	p.Arg306Cys	c 916 C>T	M
23	1,50	24,91	0,01	0,21	270,07	3	Co	p. Thr158Met	c 473 C>T	M
24	1,42	8,57	3,21	0,10	178,93	12	PSV	p.Arg294X	c.880C>T	LT
25	3,21	11,81	9,94	0,14	161,66	6	Co	p.Thr158Met	c 473 C>T	M
26	1,78	6,08	4,46	0,13	206,41	4	Co	p.Thr158Met	c 473 C>T	M
27	2,59	8,41	6,35	0,11	190,19	8	PSV	p. P389X	c 1164_1207del44	LT

Table 2: Patients Kerr
Score

Pt	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	TOT
1	0	2	1	0	1	2	2	2	1	2	0	2	1	2	2	0	1	1	2	1	25
2	1	0	2	2	2	1	1	1	1	2	0	1	1	2	2	2	1	1	0	0	23
3	2	2	2	1	1	1	2	1	2	1	0	2	2	2	2	2	2	1	1	2	31
4	0	1	1	1	1	1	1	0	1	1	0	1	1	2	2	1	1	1	0	0	17
5	0	1	2	2	2	1	1	1	1	1	0	1	0	2	2	2	1	1	1	1	23
6	0	0	2	1	1	0	0	0	1	1	0	0	1	1	1	0	1	1	0	0	11
7	1	2	0	0	1	1	1	1	2	2	1	0	1	2	2	1	1	1	0	0	19
8	0	1	1	1	1	1	0	0	2	2	0	2	1	2	2	1	1	1	1	1	21
9	1	0	2	2	1	1	1	1	1	2	0	0	1	2	1	2	1	1	2	1	23
10	0	1	1	2	1	1	1	1	0	1	0	1	0	1	1	1	1	1	1	1	17
11	1	1	1	0	0	1	1	1	0	1	0	1	0	1	1	1	0	1	1	1	14
12	1	1	2	1	1	1	0	1	1	2	0	1	2	1	2	1	0	1	2	1	22
13	1	1	1	0	1	1	1	1	1	1	0	1	0	1	1	0	0	1	0	0	13
14	2	0	1	2	0	1	0	0	1	2	0	1	2	2	2	0	1	1	0	1	19
15	1	0	1	0	0	1	1	1	1	2	1	1	1	1	2	1	2	1	0	0	18
16	0	1	1	0	1	1	1	1	1	2	0	1	0	1	0	0	1	1	0	0	14
17	1	0	2	1	1	1	0	0	1	2	0	2	0	1	2	0	1	1	0	0	16
18	0	1	2	0	1	2	0	0	2	2	0	1	1	2	2	1	1	0	0	1	19
19	0	1	0	0	0	1	0	0	2	2	0	1	1	1	2	1	0	0	0	1	13
20	2	1	1	0	0	2	0	0	1	1	0	1	1	1	1	0	0	0	0	0	12
21	0	0	0	0	0	0	1	0	1	2	0	1	1	2	2	0	0	0	0	1	11
22	0	0	0	1	0	2	0	0	1	1	0	1	0	2	1	0	0	0	1	0	10
23	0	2	1	1	1	2	0	0	2	0	1	2	2	2	2	2	2	0	1	2	25
24	0	0	0	0	2	0	1	0	1	1	0	1	0	1	1	1	0	0	2	1	12
25	1	1	2	0	0	2	1	0	2	1	0	1	0	2	2	0	0	1	0	0	16
26	1	1	1	0	0	2	1	2	2	1	0	2	1	2	2	2	1	0	0	1	22
27	0	0	0	0	0	0	1	0	0	1	1	0	0	1	1	1	0	1	1	0	8

Table 3: Patients Clinical Severity Score

Patient	age onset regression	somatic growth	head growth	motor sitting	ambulation	hand use	scoliosis	language	non verbal communication	respiratory dysfunction	autonomic symptoms	onset stereotype	epilepsy	Total CSS
1	3	0	1	2	3	2	2	3	1	1	2	2	0	22
2	3	4	u	2	1	3	2	3	2	0	1	2	4	27
3	3	0	u	2	5	3	1	3	2	1	1	3	4	28
4	3	4	u	1	1	2	0	4	0	1	1	2	3	22
5	3	2	u	1	3	3	2	4	1	1	1	3	1	25
6	3	1	u	0	1	2	1	3	1	1	1	3	0	17
7	4	4	u	2	2	2	2	3	0	1	1	2	4	27
8	4	1	u	2	5	2	1	4	2	1	1	3	0	25
9	3	0	u	1	1	3	2	3	2	2	4	3	4	28
10	4	4	u	1	1	2	2	3	0	2	1	2	1	23
11	1	0	u	0	0	1	1	1	0	1	0	1	0	6
12	4	4	u	2	1	2	3	3	2	1	1	4	0	27
13	3	0	u	1	1	0	1	2	1	0	1	3	0	13
14	2	4	u	0	0	3	0	3	1	3	1	2	0	19
15	2	4	u	2	0	3	1	3	1	2	1	2	1	22
16	2	0	u	1	2	3	0	3	2	1	1	2	0	17
17	1	1	u	0	0	1	0	4	1	1	0	1	0	10
18	2	0	4	1	5	3	0	4	1	2	0	2	0	24
19	3	0	0	5	5	2	3	4	1	0	3	3	0	29
20	0	0	0	5	5	2	0	2	2	0	0	3	0	19
21	3	0	0	1	3	3	2	3	2	0	0	3	0	20
22	3	0	0	0	1	1	0	2	2	0	0	3	0	12
23	5	0	0	5	5	3	0	4	2	2	1	0	1	28
24	2	0	0	0	0	1	2	3	1	0	0	1	0	10
25	4	3	2	2	4	3	0	4	2	2	3	3	4	36

26	5	2	2	4	5	5	1	3	3	1	2	3	0	36
27	2	0	0	1	1	2	3	2	2	2	1	1	0	17