

On-line Table: Patient demographics, genetics, and clinical scoring results

Patient ^a	Age (yr)	Sex	Mutation 1	Mutation 2	Putative Amino Acid Change 1	Putative Amino Acid Change 2	Motor Score	Visual Score	Language Score						
065-1	8.8	F	c.622C>T	c.509-1G>C	p.Arg208X	Intron mutation, splice defect	0	0	0						
455-1	3.5	M	c.509-1G>C	c.622C>T	Intron mutation, splice defect	p.Arg208X	3	3	3						
457-1	4.2	F	c.1094G>A	c.1094G>A	p.Cys365Tyr	p.Cys365Tyr	2	3	2						
	5.2						1	2	0						
	5.8						0	0	0						
	8.6						0	0	0						
	9.3						0	0	0						
457-2	10.3	F	c.1094G>A	c.1094G>A	p.Cys365Tyr	p.Cys365Tyr	0	0	0						
	5.2						1	2	0						
	5.8						0	0	0						
	8.6						0	0	0						
	9.3						0	0	0						
467-1	10.3	M	c.1439T>G	c.509-1G>C	p.Val480Gly	Intron mutation, splice defect	0	0	0						
	13.8						0	0	0						
	468-1						3.8	M	c.509-1G>C	c.622C>T	Intron mutation, splice defect	p.Arg208X	2	2	2
	4.7						1	1	0						
	5.6						0	0	0						
473-1	6.6	F	c.311T>A	c.509-1G>C	p.Leu104X	Intron mutation, splice defect	0	0	0						
	7.9						0	0	0						
	5.7						1	3	1						
	6.3						0	0	0						
	7.2						0	0	0						
479-1	2.3	F	c.622C>T	c.622C>T	p.Arg208X	p.Arg208X	2	2	2						
	7.1	0	1	0											
481-1	6.4	F	c.622C>T	c.622C>T	p.Arg208X	p.Arg208X	0	0	0						
486-1	3.3	F	c.509-1G>C	c.833A>G	Intron mutation, splice defect		3	3	3						
495-1	5.2	M	c.380G>A	c.509-1G>C	p.Arg127Gln	Intron mutation, splice defect	3	2	2						
499-1	6.5	M	c.230-12T>A	c.622C>T	Intron mutation	p.Arg208X	0	2	0						
532-1	3.1	F	c.622C>T	c.622C>T	p.Arg208X	p.Arg208X	3	3	3						

^a Patients 0457-1 and 0457-2 are identical twins.