

Table S5. Major clinical features and MARS2 mutations in French Canadian individuals with ARSAL

Patient no.	Gender	Family	Mutation 1	Mutation 2	Mutation detection	Age of onset	Ataxia	Spasticity	Dysarthria	Dysmetria	Nystagmus	Dystonia	Scoliosis	U. urgency	Wheelchair use, y	leukodystrophy	Cerebellar atrophy			
																	Vermis	Hemispheres		
10	M	E	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays, CGH-array, Western Blot	15	*	*	*	*	*	*	*	*			+	++	++	
11	M	E	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays, Western Blot	3	*	*	*	*	*	*	*	*						
3	F	B	Dup1	Dup1	PCR, Sequencing, CNV assays, CGH-array, Western Blot	3	*	*	*	*	*	*	*	*	*	51		+	+	+
4	M	B	Dup1	Dup1	PCR, Sequencing, CNV assays, CGH-array, Western Blot	6	*	*	*	*	*	*	*	*	*	19		+	+++	+++
5	M	B	Dup1	Dup1	PCR, Sequencing, CNV assays, Western Blot	3	*	*	*	*	*	*	*	*	*	45		+	++	+
6	M	C	Dup2	Dup2	PCR, Sequencing, CNV assays, Western Blot	20	*	*	*	*	*	*	*	*	*	59		+	++	++
8	F	C	Dup2	Dup2	PCR, Sequencing, CNV assays, Western Blot	17	*	*	*	*	*	*	*	*	*			-	++	++
1	F	C	Dup2	Dup2	PCR, Sequencing, CNV assays	47	*	*	*	*	*	*	*	*	*					
2	M	C	Dup2	Dup2	PCR, Sequencing, CNV assays	50	*	*	*	*	*	*	*	*	*					
9	F	D	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays, Western Blot	13	*	*	*	*	*	*	*	*	*	30		-	+	+
12	F	A	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays	3	*	*	*	*	*	*	*	*	*	10		-	+++	+++
13	F	A	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays, CGH-array	3	*	*	*	*	*	*	*	*	*			-	+++	+++
14	M	F	Dup1	Dup1	PCR, Sequencing, CNV assays	5	*	*	*	*	*	*	*	*	*			+	+	-
15	F	G	Dup1	Dup1	PCR, Sequencing, CNV assays	3	*	*	*	*	*	*	*	*	*	45		-	+++	+++
7	M	H	Dup1	Dup1	PCR, Sequencing, CNV assays	19	*	*	*	*	*	*	*	*	*			-	+++	+++
17	M	I	Dup1	Dup1	PCR, Sequencing, CNV assays	23	*	*	*	*	*	*	*	*	*			-	+++	+++
18	M	I	Dup1	Dup1	PCR, Sequencing, CNV assays	30	*	*	*	*	*	*	*	*	*	38		-	+++	++
16	F	J	Dup1	Dup2	PCR, Sequencing, CNV assays	2	*	*	*	*	*	*	*	*	*	36		-	+	-
21	F	K	Dup1	Dup1	PCR, Sequencing, CNV assays	59	*	*	*	*	*	*	*	*	*	72		-	+	+
18	F	L	Dup1	Dup2	PCR, Sequencing, CNV assays	43	*	*	*	*	*	*	*	*	*			-	++	++
19	M	M	Dup1	Dup2	PCR, Sequencing, CNV assays	26	*	*	*	*	*	*	*	*	*	38		+	++	++
20	M	N	Dup1	Dup2	PCR, Sequencing, CNV assays	4	*	*	*	*	*	*	*	*	*			-	+++	+++
24	M	P	Dup1	Dup1	PCR, Sequencing, CNV assays, Western Blot	9	*	*	*	*	*	*	*	*	*	16		+	+	-
26	F	R	Dup1	Dup1	PCR, Sequencing, CNV assays	2	*	*	*	*	*	*	*	*	*			-	+++	+++
28	F	T	Dup1	Dup1	PCR, Sequencing, CNV assays	32	*	*	*	*	*	*	*	*	*			+	+++	+++
29	M	U	Dup1	Dup1	PCR, Sequencing, CNV assays	48	*	*	*	*	*	*	*	*	*			-	+	+
30	F	X	Dup1	Dup1	PCR, Sequencing, CNV assays	50	*	*	*	*	*	*	*	*	*			-	+	-
31	M	Y	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays	5	*	*	*	*	*	*	*	*	*	14		-	+	-
32	M	Z	Dup1	Dup1	PCR, Sequencing, CNV assays	13	*	*	*	*	*	*	*	*	*			-	+++	+++
33	F	Z	Dup1	Dup1	PCR, Sequencing, CNV assays	48	*	*	*	*	*	*	*	*	*			+	+	+
34	M	AA	Dup1	Dup2	PCR, Sequencing, CNV assays	29	*	*	*	*	*	*	*	*	*			-	+	+
35	F	AA	Dup1	Dup2	PCR, Sequencing, CNV assays, Western Blot	26	*	*	*	*	*	*	*	*	*			-	++	++
36	M	BB	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays	16	*	*	*	*	*	*	*	*	*			-	+	+
37	M	CC	Dup1	Dup1	PCR, Sequencing, CNV assays	28	*	*	*	*	*	*	*	*	*	53		-	+++	++
38	M	DD	Dup1	Dup2	PCR, Sequencing, CNV assays	36	*	*	*	*	*	*	*	*	*			-	+	+
39	M	DD	Dup1	Dup2	PCR, Sequencing, CNV assays, Western Blot	40	*	*	*	*	*	*	*	*	*			-	-	-
40	F	DD	Dup1	Dup2	PCR, Sequencing, CNV assays, Western Blot	35	*	*	*	*	*	*	*	*	*			-	++	++
41	M	EE	Dup1	Dup1	PCR, Sequencing, CNV assays, Western Blot	25	*	*	*	*	*	*	*	*	*			-	-	-
42	M	FF	Dup1	Dup1	PCR, Sequencing, CNV assays	30	*	*	*	*	*	*	*	*	*			-	+	+
43	F	GG	Dup2	Dup2	PCR, Sequencing, CNV assays	30	*	*	*	*	*	*	*	*	*			+	++	++
44	F	HH	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays, Western Blot	13	*	*	*	*	*	*	*	*	*			+	+	-
45	M	HH	Dup1	c.681Δ268bpfx236X	PCR, Sequencing, CNV assays, Western Blot	14	*	*	*	*	*	*	*	*	*			+	-	-
46	M	II	Dup2	Dup2	PCR, Sequencing, CNV assays	32	*	*	*	*	*	*	*	*	*			-	-	-
47	M	II	Dup2	Dup2	PCR, Sequencing, CNV assays	44	*	*	*	*	*	*	*	*	*			-	-	-
48	M	JJ	Dup1	Dup1	PCR, Sequencing, CNV assays	41	*	*	*	*	*	*	*	*	*			+	-	-
49	F	KK	Dup1	Dup2	PCR, Sequencing, CNV assays	37	*	*	*	*	*	*	*	*	*			-	+++	++
50	M	KK	Dup1	Dup2	PCR, Sequencing, CNV assays	20	*	*	*	*	*	*	*	*	*	42		-	+++	+++
51	F	KK	Dup1	Dup2	PCR, Sequencing, CNV assays	38	*	*	*	*	*	*	*	*	*			+	+	+
52	M	LL	Dup1	Dup1	PCR, Sequencing, CNV assays	13	*	*	*	*	*	*	*	*	*			+	+	+
53	F	MM	Dup1	Dup1	PCR, Sequencing, CNV assays	40	*	*	*	*	*	*	*	*	*			-	+++	+++
54	M	NN	Dup1	Dup2	PCR, Sequencing, CNV assays	40	*	*	*	*	*	*	*	*	*	51		+	+++	-
55	F	OO	Dup1	Dup1	PCR, Sequencing, CNV assays	45	*	*	*	*	*	*	*	*	*	59		-	++	++
56	F	PP	Dup1	Dup1	PCR, Sequencing, CNV assays	2	*	*	*	*	*	*	*	*	*			-	+++	-
57	F	RR	Dup1	Dup1	PCR, Sequencing, CNV assays	40	*	*	*	*	*	*	*	*	*			-	+	+
Mean/%						24.4	100%	100%	78%	57%	44%	28%	17%	48%	39.9		40%	93%	75%	

Legend:
Dup1: Duplication 1; Dup2: Duplication 2; U. urgency: urinary urgency; y: year; CNV: Copy number variation; CGH: copy genomic hybridization