Supplementary Table 1. *NEU1* variants associated with sialidosis type 1

AA change (A1)	AA change (A2)	Age at onset (yr)	Gender	Ataxia	Dysarthria	Myoclonus	Seizure	Impaired cognition	Cherry-red spot	Abnormal EEG	Abnormal SEP	Abnormal VEP	Abnormal MRI	Reference
p.V143E	p.S182G	13	F	Y	Y	Y	Y	N	Y	Y	N	Y	N	This study
		14	M	Y	Y	Y	Y	N	Y	N	NA	Y	N	This study
p.P80L	p.Y268C	12	M	Y	Y	Y	Y	N	Y	NA	Y	Y	N	1
p.P80L	p.S182G	12	F	Y	Y	Y	Y	Y	Y	NA	Y	Y	Y	1
		17	M	Y	Y	Y	Y	N	Y	NA	Y	N	N	1
		8	M	N	N	N	N	N	N	N	NA	Y	N	2
		12	F	Y	Y	Y	Y	N	Y	Y	NA	Y	N	2
		17	M	Y	NA	Y	Y	Y	NA	NA	NA	NA	NA	3
p.R280X	p.D135N	10	F	Y	Y	Y	Y	N	Y	NA	NA	NA	N	1
		10	F	Y	Y	Y	Y	N	Y	N	NA	Y	N	2
c.1118T>C	p.S182G	10	M	Y	Y	Y	Y	N	Y	NA	Y	Y	Y	1
p.R341X	p.S182G	10	M	Y	Y	Y	Y	N	Y	Y	NA	NA	N	2
p.S182G	p.A106_G118del	12	M	Y	N	Y	Y	N	N	Y	Y	Y	N	4
p.S182G	p.S182G	14	M	Y	Y	N	Y	NA	NA	NA	NA	NA	Y	5
		NA	M	NA	Y	Y	NA	NA	NA	NA	NA	NA	NA	6
		27	M	Y	NA	Y	Y	N	N	N	NA	NA	N	7,8
		19	M	Y	NA	Y	Y	NA	N	Y	NA	NA	Y	7,8
		14	M	Y	NA	Y	Y	N	N	NA	NA	NA	NA	7
		26	M	Y	NA	Y	N	N	N	N	NA	NA	Y	7,8
		16	F	Y	NA	N	Y	N	N	NA	NA	NA	NA	7
		12	M	Y	NA	N	Y	N	N	NA	NA	NA	NA	7
		20	F	Y	NA	Y	Y	N	N	N	NA	NA	Y	7,8
		33	M	Y	NA	Y	Y	N	N	N	NA	NA	Y	7,8
		20	M	Y	NA	Y	N	N	N	N	NA	NA	N	7,8
		15	M	Y	NA	Y	N	N	N	N	NA	NA	Y	7,8
		18	M	Y	NA	Y	Y	N	N	NA	NA	NA	NA	7
		28	F	Y	NA	Y	Y	N	N	NA	NA	NA	NA	7
		19	M	Y	NA	Y	N	N	N	N	NA	NA	Y	7,8
		18	F	Y	NA	Y	N	N	N	N	NA	NA	N	7,8
		14	M	Y	NA	Y	Y	N	Y	N	NA	NA	Y	7,8
p.G328S	p.L91R	13	F	Y	Y	Y	Y	NA	Y	Y	Y	Y	Y	9
p.G328S	p.S403Tfs*85	13	F	Y	Y	Y	Y	N	N	Y	Y	NA	Y	9,10
		12	F	Y	N	Y	N	N	Y	Y	Y	NA	Y	9
p.R305H	p.E209Sfs*94	17	F	Y	Y	Y	Y	N	N	Y	N	N	Y	9,10
p.S182G	27.5 kb deletion	9	F	N	N	N	N	N	Y	NA	NA	NA	N	11
p.D310N	p.P6Qfs*21	12	F	Y	Y	Y	Y	N	Y	Y	NA	NA	Y	12
p.R280X	g.2869A>T	14	F	Y	N	Y	N	N	Y	N	Y	Y	Y	13
p.S182G	c.667_679del	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	14
p.P210L	p.P210L	16	M	Y	N	Y	Y	NA	Y	Y	NA	Y	N	15
p.S182G	p.G227R	12	M	Y	Y	Y	Y	N	N	N	NA	NA	N	16
p.Glu209SerfsTer94	p.D310N	18	M	Y	Y	Y	Y	NA	Y	NA	NA	NA	Y	17
p.S182G	c.619C>T	12	M	Y	Y	Y	Y	N	Y	Y	NA	NA	Y	18,19
p.G248C	p.G248C	9	M	Y	N	Y	N	N	Y	N	NA	NA	N	20
p.S233R	p.Y268C	8	M	N	N	N	N	N	Y	Y	NA	Y	N	21
p.G227R	p.H399_Y400dup	12	F	Y	N	N	N	N	Y	NA	NA	NA	N	22
		13	F	Y	N	Y	N	N	Y	NA	NA	NA	N	22
p.G328S	p.G227R	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	10
p.D234N	p.R341X	26	M	Y	Y	Y	N	NA	Y	Y	NA	NA	Y	23
		NA	NA	NA	NA	Y	NA	NA	Y	NA	NA	NA	NA	24
p.S67I	p.S67I	23	M	N	N	Y	N	N	N	N	Y	NA	N	25
		25	F	Y	N	Y	N	N	N	N	Y	NA	Y	25
		22	F	Y	N	Y	Y	N	N	N	Y	NA	Y	25
p.G227R	p.R305C	25	F	N	N	Y	N	N	N	N	Y	NA	Y	25
		25	F	N	N	Y	N	N	N	N	Y	NA	N	25
		32	M	N	N	Y	N	N	N	N	Y	NA	N	25
p.P80L	p.D135N	14	M	Y	Y	Y	N	Y	Y	NA	NA	NA	Y	26
p.Q282H	p.Q282H	13	NA	Y	N	Y	N	N	N	NA	NA	NA	NA	24
p.R294C	c.1191delG	14	F	Y	Y	Y	N	N	Y	NA	NA	NA	NA	27
p.V217M	p.G243R	15	M	Y	N	Y	Y	Y	Y	N	Y	NA	N	28
		17	F	Y	Y	N	Y	N	Y	NA	NA	NA	NA	29
		32	M	Y	Y	Y	N	N	Y	NA	NA	NA	NA	29
p.S182G	p.Q55stop	12	F	Y	N	Y	Y	N	Y	NA	NA	NA	NA	7,8
p.S182G	p.A319V	14	M	Y	N	Y	Y	N	Y	N	NA	NA	N	7,8
p.G136E	p.V275A	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	30
p.L111P	ND	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	30
p.T345I	p.T345I	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	30
p.P316S	p.P316S	14	M	Y	Y	Y	Y	N	Y	NA	NA	NA	NA	31
p.R294S	p.L231H	11	F	Y	Y	Y	Y	N	Y	NA	NA	NA	NA	32
p.R294S	p.C218A	8	F	N	N	N	N	Y	Y	NA	NA	NA	NA	32
p.V54M	p.G378stop	17	F	Y	Y	Y	Y	N	Y	NA	NA	NA	NA	32
		15	F	Y	Y	Y	Y	N	N	NA	NA	NA	NA	32
p.G328S	p.Dpl399HisTyr	13	F	Y	Y	Y	Y	N	N	Y	NA	Y	NA	32
p.V54M	p.E377X	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	33

AA, amino acid; EEG, electroencephalography; SEP, somatosensory-evoked potential; VEP, visual evoked potential; MRI, magnetic resonance imaging; F, female; M, male; Y, yes; N, no; NA, not applicable; ND, not determined.

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