

Subject ID	Inheritance	cDNA	Sex	Age	MRI	EEG Results	Medications at Time of EEG	Biopsy
1	de novo	c.340C>T	M	2	Age 10 mo: Delayed myelination, restricted diffusion bilateral tegmental tracts	Not Done	n/a	none
2	de novo	c.340C>T	F	4	Not Done	Not Done	n/a	none
3	de novo	c.616C>T	F	19	Ages 18 mo and 9 yo: normal	Age 18 mo: normal	none	none
4	de novo	c.616C>T	F	8	Normal (not available for review)	Age 4 yo: Abnormal due to (1) 7 Hz PDR, (2) Diffuse background slowing, most notable in posterior quadrants, (3) Burst of generalized spike-wave discharges were rarely seen during sleep, (4) No seizures Age 7 yo: Abnormal due to (1) No defined PDR, (2) Diffuse beta throughout the recording, maximal in the bilateral frontal regions, Diffusely slow background for Age, predominately delta and theta frequencies, (3) No epileptiform discharges, (4) No seizures	none	none
5	de novo	c.616C>T	F	3	Age 10 mo: Delayed myelination	Age 9 mo normal	none	none
6	de novo	c.616C>T	M	17	Ages 8 mo and 14 mo: Delayed myelination	Age 3 yo: normal	none	none
7	de novo	c.616C>T	F	38	Age 10 yo: normal	Age 37 yo: normal	none	none
8	de novo	c.616C>T	F	11	Age 3 yo: normal	Age 5 yo: Abnormal activity occipito-temporal regions	none	Age 10 mo: increased small myofibers with mild myofiber size variation; mild type II fiber atrophy by muscle enzyme histochemistry with mild increase in morphologically normal glycogen & mitochondria
9	de novo	c.616C>T	F	18	Ages 1 yo and 2 yo: Normal; 5 yo MRI/MRS with Elevated lactate peak in basal ganglia	Age 4 yo: Bilateral occipito-temporal epileptiform discharges; Age 12: diffuse slowing	none	none
10	de novo	c.616C>T	F	6	Age 1 yo: normal	Age 3 yo 11 mo: normal	none	Age 3 yo: normal
11	de novo	c.616C>T	F	13	Age 1 yo: normal	Age 1 yo: normal	none	none
12	de novo	c.616C>T	F	10	Age 2 yo: normal	Not Done	n/a	none
13	de novo	c.616C>T	F	27	Not Done	Not Done	n/a	none
14	de novo	c.616C>T	F	3	Age 17 mo: normal	Age 2 yo: normal	none	none
15	de novo	c.616C>T	F	8	Age 12 mo: normal	Age 7 yo: normal	none	none
16	de novo	c.616C>T	F	17	Age 2 yo: normal	Ages 2 yo, 6 yo, 15 yo: normal	none	none
17	de novo	c.616C>T	F	23	Age 11 yo: normal	Not Done	n/a	none
18	de novo	c.616C>T	F	24	Age 5 mo, 2 yo, 5 yo: normal; MRS age 5 yo: normal	Age 3 yo: Mild diffuse slowing with overabundance of faster beta activity; Age 22 yo: mild diffuse slowing	PIR	Age 6 yo: normal
19	de novo	c.616C>T	F	28	Not Done	Not Done	n/a	none
20	de novo	c.616C>T	F	6	Age 4 yo: normal	Age 4 yo: normal	none	none
21	de novo	c.616C>G	F	20	Not Done	Age 10 yo: normal	none	none
22	de novo	c.617G>A	F	36	Age 34 yo: Multiple acute infarctions of the left frontal, parietal and temporal lobes. Likely sub-acute infarcts of left basal ganglia and left cerebellum. Vessel imaging unremarkable.	Age 9 mo: normal	none	none
23	de novo	c.617G>A	F	8	Age 14 mo: normal	Age 6 yo: abnormal (results not available)	none	none
24	de novo	c.617G>A	M	17	Age 12 yo: normal	Age 12 yo: left hemisphere slowing	none	Age 2 yo: normal
25	de novo	c.617G>A	F	17	Age 2 yo, 6 yo, 8 yo: normal	Normal (clinician reported)	none	none
26	de novo	c.617G>A	F	11	Age 12 mo: Normal	Routine EEG: Very rare epileptiform discharges in bilateral rolandic regions	none	none
27	de novo	c.617G>T	F	36	Age 32 yo: Hypoplasia of the inferior cerebellar vermis, slightly atrophic folia of the superior vermis	Age 7 yo: normal	none	none
28	de novo	c.626C>T	F	10	Age 3 yo: normal	Ages 2 yo, 3 yo: normal	LVT	none
29	de novo	c.629A>G	F	5	Age 1 yo: normal	Not Done	n/a	none
30	de novo	c.634A>G	F	5	Age 3 yo: normal	Age 4 yo: sharp and spike wave left frontoparietal	SUL	Age 4 mo: Normal histologically with reduced activity complex II and III of the respiratory chain enzymes
31	de novo	c.635G>C	F	8	Ages 18 mo and 5 yo: normal	Age 5 yo: mild background slowing, occasional centrotemporal R>L	none	none
32	de novo	c.638C>T	F	20	Not Done	Not Done	n/a	none
33	de novo	c.1019A>T	M	8	Ages 1.5 yo and 3 yo: normal	Age 6 yo: Left posterior and midline epileptiform discharges	none	none

**Supplemental Table 2: Additional Testing including MRI, EEG and Muscle Biopsy Results**

LVT = levetiracetam, SUL = sulthiame