

Supplementary Table 1: Clinical and genetic data of patient cohort

Patient ID	Sex	MLD classification	Age at MRI (y)	Age at onset (y)	GMFC-MLD	Mutation 1		Mutation 2		FSIQ
						cDNA level	protein level	cDNA level	protein level	
MLD1	m	Adult	28	27	0	c.251G>A	p.Arg84Gln	c.287C>T	p.Ser96Phe	72
MLD2	f	Juvenile	8	8	1	c.459+1G>A	p.?	c.1277C>T	p.Pro426Leu	65
MLD3	m	Juvenile	7	5	1	c.459+1G>A	p.?	c.1277C>T	p.Pro426Leu	71
MLD4	f	Juvenile	6	5	1	c.634G>C	p.Ala212Pro	c.1277C>T	p.Pro426Leu	68
MLD5	f	Juvenile	14	14	1	c.1277C>T	p.Pro426Leu	c.1277C>T	p.Pro426Leu	94
MLD6	f	Adult	35	35	0	c.251G>A	p.Arg84Gln	c.287>C>T	p.Ser96Phe	87
MLD7	f	Juvenile	6	n.a.	0	c.459+1G>A	p.?	c.536T>G	p.Ile179Ser	n.a.
MLD8	m	Adult	17	17	0	c.635C>T	p.Ala212Val	c.1277C>T	p.Pro426Leu	100
MLD9	f	Juvenile	15	14	1	c.635C>T	p.Ala212Val	c.1277C>T	p.Pro426Leu	65
MLD10	m	Late-infantile	2	1.5	6	c.1069C>T	p.Gln232*	c.1216G>T	p.Asp281Tyr	n.a.
MLD11	f	Juvenile	7	6	1	c.1277C>T	p.Pro426Leu	c.1372C>T	p.Gln458*	61
MLD12	f	Adult	26	30	0	c.536T>G	p.Ile170Ser	c.1171A>G	p.Ser391Gly	n.a.
MLD13	f	Juvenile	6	4	5	n.a.		n.a.		n.a.
MLD14	f	Adult	25	25	0	c.459+1G>A	p.?	c.536T>G	p.Ile179Ser	91
MLD15	f	Late-infantile	2	1.6	2	c.459+1G>A	p.?	c.830_831delTCinsAA	p.Ile277Lys	101 ⁺⁺
MLD16	m	Late-infantile	2	1.3	2 ⁺	c.245C>T	p.Pro82Leu	c.1168C>T	p.Arg390Trp	111 ⁺⁺
MLD17	f	Juvenile	7	5	1	c.830_831delTCinsAA	p.Ile277Lys	c.1277C>T	p.Pro426Leu	74
MLD18	f	Juvenile	12	11	1	c.1277C>T	p.Pro426Leu	c.1277C>T	p.Pro426Leu	n.a.
MLD19	f	Adult	32	27	0	c.571C>A	p.Pro191Thr	c.1277C>T	p.Pro426Leu	53
MLD20	f	Juvenile	7	6	1	c.459+1G>A	p.?	c.1277C>T	p.Pro426Leu	63

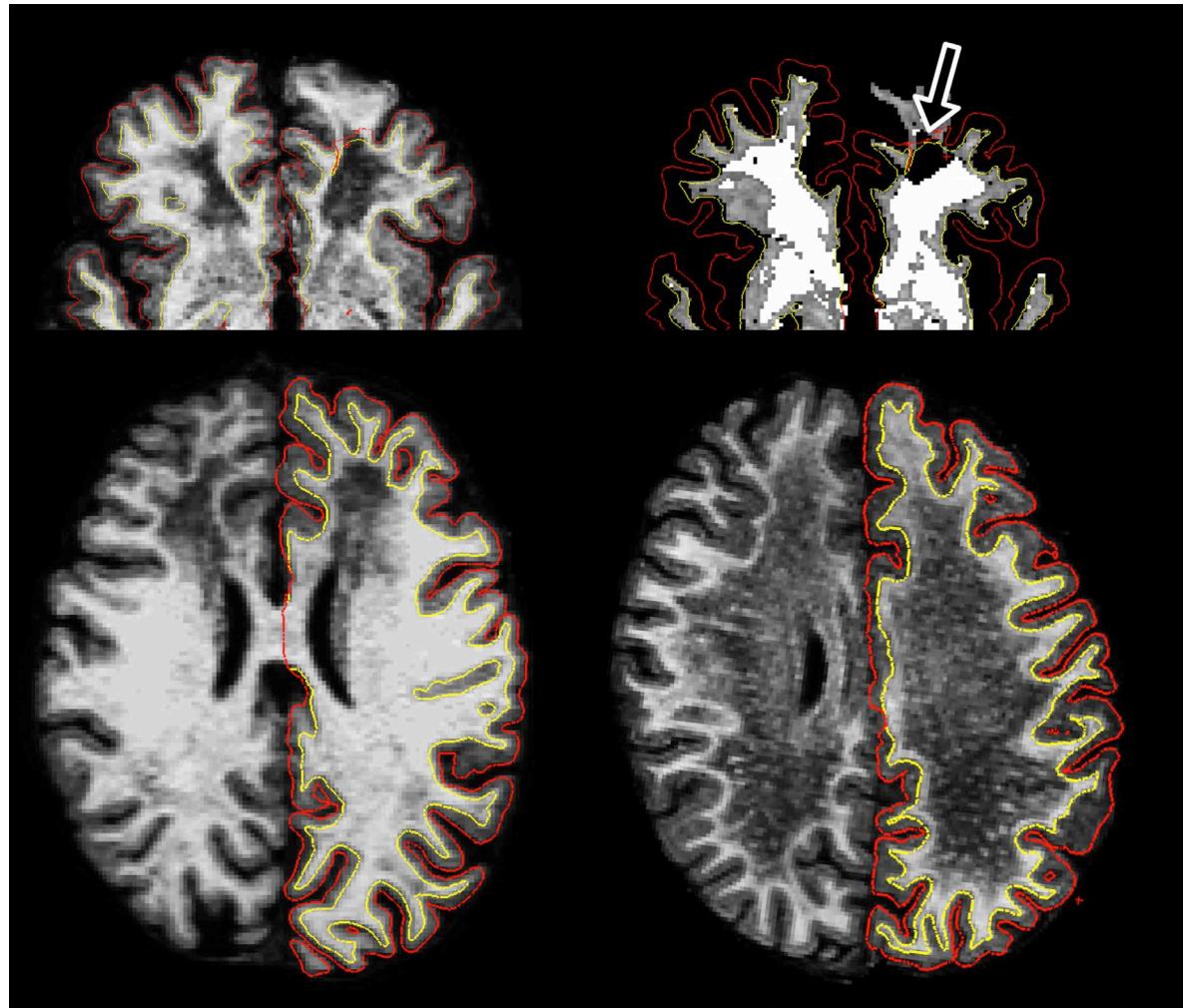
[†] motor problems secondary to severe demyelinating polyneuropathy

⁺⁺ developmental index

FSIQ: Full Scale IQ

n.a. not available

Supplementary Figure 1: Correction method in MLD



Examples of correction method in MLD patients. In the upper panel, an example is shown where the initial processing resulted in significant errors in the frontal lobe. On the upper right hand image the area not recognized as WM (arrow) was filled and subsequent segmentation was without difficulty. In the bottom panel, two examples are provided after this filling method, showing successful outlining of the left hemisphere surfaces in MLD patients. (yellow – white surface, red – pial surface).