Table e-1. Overview of the levels of the GMFCS, MACS and CFCS[1]

GMFCS: Gross Motor Function Classification System[2]
MACS: Manual Ability Classification System[3]
CFCS: Communication Function Classification System[1]

	GMFCS **Motor function**	MACS **Manual ability**	CFCS **Communication**
LEVEL I	Walks without limitations	Handles objects easily and successfully	Sends and receives with familiar and unfamiliar partners effectively and efficiently
LEVEL II	Walks with limitations	Handles most objects but with somewhat reduced quality and/or speed of achievement	Sends and receives with familiar and unfamiliar partners but may need extra time
LEVEL III	Walks using a hand-held mobility device	Handles objects with difficulty; needs help to prepare and/or modify activities	Sends and receives with familiar partners effectively, but not with unfamiliar partners
LEVEL IV	Self-Mobility with limitations; may use powered mobility	Handles a limited selection of easily managed objects in adapted situations	Inconsistently sends and／or receives even with familiar partners
LEVEL V	Transported in a manual wheelchair	Does not handle objects and has severely limited ability to perform even simple actions	Seldom effectively sends and receives, even with familiar partners

Table e-2. Genotype-phenotype correlation

MLC1 mutations	Number of informative patients (families)	Median age at phenotyping [range]	Loss of ambulation[1]	Wheelchair dependency[1]	Seizures[2]
c.135insC, p.Cys46Leufs*34 homozygous	4 (4)	26 [14 -45] y	(a) (b) (d)	(a) (b)	(a) (c)
c.268_422del, p.Cys90_Ile141del homozygous	4 (3)	14 [10 - 20] y	(a) (d)	(a) (c) (d)	(b) (c)
c.278C>T, p.Ser93Leu homozygous	4 (4)	31 [14 - 39] y	(a) (b) (c)	(a) (b) (c)	(c) (d)
c.353C>G, p.Thr118Met homozygous	4 (3)	18 [12 - 24] y	(a) (c)	(a)	(a) (c)
c.424-3C>G, p.? homozygous	4 (2)	24 [19 - 27] y	(b) (c)	(a) (b)	(c) (d)
c.908_918delinsGCA, p.Val303Glyfs*96 homozygous	5 (3)	16 [12 - 20] y	(a) (b)	(a)	(a) (b) (c)

y, years

[1] **(a)**: function not lost **(b)**: Loss at age ≥ 11 years **(c)**: loss at age 6-10 years; **(d)**: loss at age 0-5 years

[2] **(a)**: absent; **(b)**: single seizure; **(c):** well controlled epilepsy; **(d)**: moderately controlled/refractory epilepsy

Table e-3. Characteristics first MRI

AGE AT MRI	MLC1		MLC2A		MLC2B	
	<2 years	**≥ 2 years**	**<2 years**	**≥ 2 years**	**<2 years**	**≥ 2 years**
Number of patients	50	91	5	11	28	2
CEREBRAL WM						
Signal PLIC						
normal	0% (0/49)	0% (0/89)	0% (0/5)	0% (0/11)	54% (15/28)	50% (1/2)
posterior part abnormal	2% (1/49)	7% (6/89)	20% (1/5)	9% (1/11)	14% (4/28)	50% (1/2)
double line throughout	92% (45/49)	93% (83/89)	80% (4/5)	91% (10/11)	32% (9/28)	0% (0/2)
abnormal throughout	6% (3/49)	0% (0/89)	0% (0/5)	0% (0/11)	0% (0/28)	0% (0/2)
Abnormal signal ALIC	92% (45/49)	21% (19/89)	60% (3/5)	10% (1/10)	29% (8/28)	0% (0/2)
Sparing subcortical WM						
frontal	13% (6/47)	27% (22/81)	40% (2/5)	20% (2/10)	11% (3/28)	50% (1/2)
temporal	13% (6/47)	26% (21/81)	80% (4/5)	20% (2/10)	25% (7/28)	100% (2/2)
parietal	21% (10/47)	61% (49/81)	80% (4/5)	50% (5/10)	36% (10/28)	100% (2/2)
occipital	38% (18/47)	84% (68/81)	80% (4/5)	80% (8/10)	57% (16/28)	100% (2/2)
Sparing of WM in certain brain area(s)						
frontal	0% (0/49)	6% (5/78)	0% (0/5)	0% (0/10)	0% (0/28)	50% (1/2)
temporal	0% (0/49)	3% (2/78)	0% (0/5)	0% (0/10)	4% (1/28)	50% (1/2)
parietal	0% (0/49)	6% (5/78)	0% (0/5)	0% (0/10)	14% (4/28)	100% (2/2)
occipital	4% (2/49)	12% (9/78)	20% (1/5)	10% (1/10)	11% (3/28)	100% (2/2)
Widening ventricles	6% (3/50)	25% (23/91)	0% (0/5)	27% (3/11)	7% (2/28)	50% (1/2)
Enlargement SAS	12% (6/50)	34% (31/91)	0% (0/5)	36% (4/11)	46% (13/28)	50% (1/2)
CYSTS AND NEAR-CYSTIC RAREFACTION OF THE WM						
Number of cysts						
0	0% (0/47)	0% (0/85)	40% (2/5)	10% (1/10)	43% (12/28)	50% (1/2)
1 - 2	64% (30/47)	36% (31/85)	20% (1/5)	30% (3/10)	36% (10/28)	50% (1/2)
3 - 6	34% (16/47)	52% (44/85)	20% (1/5)	40% (4/10)	21% (6/28)	0% (0/2)
> 6	2% (1/47)	12% (10/85)	20% (1/5)	20% (2/10)	0% (0/28)	0% (0/2)
Location of cysts						
frontal	30% (14/47)	54% (46/85)	40% (2/5)	60% (6/10)	21% (6/28)	0% (0/2)
temporal	98% (46/47)	99% (84/85)	60% (3/5)	90% (9/10)	57% (16/28)	50% (1/2)
parietal	17% (8/47)	37% (31/85)	20% (1/5)	20% (2/10)	7% (2/28)	0% (0/2)
occipital	0% (0/47)	0% (0/85)	0% (0/5)	0% (0/10)	0% (0/28)	0% (0/2)
Near cystic WM rarefaction	88% (42/48)	93% (79/85)	60% (3/5)	100% (11/11)	96% (27/28)	100% (2/2)
Overall quantity of cystic/near cystic WM						
none or little	6% (3/50)	93% (79/90)	40% (2/5)	18% (2/11)	32% (9/28)	50% (1/2)
moderate	88% (44/50)	93% (79/90)	40% (2/5)	64% (7/11)	64% (18/28)	50% (1/2)
large	6% (3/50)	93% (79/90)	20% (1/5)	18% (2/11)	4% (1/28)	0% (0/2)
Cavum septi pellucidi	100% (50/50)	100% (90/90)	100% (5/5)	100% (11/11)	96% (27/28)	100% (2/2)
Cavum vergae	69% (34/49)	91% (79/87)	100% (0/5)	100% (11/11)	82% (23/28)	100% (2/2)
INFRATENTORIAL FINDINGS						
Signal abn. cerebellar WM	96% (47/49)	78% (69/88)	80% (4/5)	90% (9/10)	18%* (5/28)	0% (0/2)
Signal abn. brainstem	96% (45/47)	95% (80/84)	100% (5/5)	91% (10/11)	86% (24/28)	100% (2/2)

abn., abnormalities; WM, white matter; PLIC, posterior limb of the internal capsule; ALIC, anterior limb of the internal capsule; SAS, subarachnoid space; * subtle T_2-hyperintense rim of the subcortical white matter, consistent with normal myelination stage up to ~5 months

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