

Supplementary Table 1. Main neurologic and MRI findings

Pt #	Neurologic and Developmental Findings	MRI findings
1	Developmental delay and hypotonia	Normal
2	Developmental delay and hypotonia, seizures, encephalopathy	Severe supratentorial, inferior cerebellar and vermis volume loss. Demyelination of the remaining supratentorial white matter and deep gray nuclei. Enlargement of the lateral and third ventricles with thinning of the corpus callosum and brainstem.
3	Developmental delay	NP
4	Normal	NP
5	Developmental delay	Multiple foci of T1 and T2 shortening in the subcortical white matter of the bilateral frontal and parietal lobes and the left temporal lobe. Several of these foci demonstrate restricted diffusion and signal loss on susceptibility weighted imaging. Imaging appearance in keeping with white matter injury in the newborn.
6	Developmental delay	Normal
7	Developmental delay	NP
8	Developmental delay	Bilateral focal periventricular defect between corpus and tail of caudate nucleus; mild, heterogenous hyperintensity of peritrigonal white matter on T2/Flair; limited cerebral and cerebellar volume loss
9	Normal	Normal
10	Developmental delay	Mild prominence of ventricles sulci.
11	Developmental delay; autism spectrum disorder	Mild prominence of the ventricular system with accompanying prominence of the frontoparietal extra-axial CSF spaces. Benign external hydrocephalus vs mild communicating hydrocephalus.

NP: not performed

Supplementary Table 2. Summary of laboratory findings.

Pt #	Age (range, years)	T HcY*	MMA*	Met*
1	0.1 - 1.8	47.2 ± 14.6	4129.7 ± 6385.1	31.0 ± 22.2
2	0.1 - 1.9	43.6 ± 22.1	6534.7 ± 5738	17.5 ± 12.8
3	0.1 - 4.8	45.5 ± 16.8	12202.8 ± 16116.6	78.6 ± 72.8
4	0.2 - 2.8	13.3 ± 2.1	1838.9 ± 1726.7	23.8 ± 11.2
5	0.1 - 4.2	48.9 ± 19.2	6752.7 ± 5708.3	18.8 ± 15.9
6	0.1 - 3.7	67.5 ± 16.9	8464.6 ± 8534.9	30.8 ± 41.6
7	4.7 - 9.3	58.8 ± 10.5	16890.3 ± 11779.6	28.4 ± 34.8
8	na	na	na	na
9	6.1 - 10.8	39.6 ± 7.0	8240.8 ± 3973.4	30.3 ± 12
10	10.7 - 15.1	143.4 ± 11.3	23793.6 ± 5250.0	35.7 ± 24.2
11	4.7 - 15.9	89.8 ± 16	25934 ± 6465.5	32.2 ± 21.3

*Values are expressed as mean ± SD

na, not available; T HcY, total plasma homocystein; MMA, plasma methylmalonic acid; Met, plasma methionine.
Reference range and units used: T HcY: 0.8 - 6.5 umol/L; MMA: 100 - 370 nmol/L; Met: 8 – 49 nmol/ml.