

Table S1. Clinical features of PMS patients compared to the subjects in this study

Features	PMS patients*	Our study
Mean age at diagnosis		11.2 years
Born at term		85%
Born pre-term		15%
Walking		Mean age 24 months
Neurologic		
Developmental delay	>95%	100%
Delayed/absent language	>95%	100%
Neonatal hypotonia	>95%	77% (still present, 41%)
Growth		
Normal		69%
Accelerated		31% ^a
Seizures		
Clinical epileptic seizures	>25%	30%
Abnormal EEG epileptic anomalies		27%
Abnormal EEG with focal or diffused slow abnormalities		32%
Facial dysmorphisms		
Long eyelashes	>75%	55%
Dolichocephaly	>50%	18%
Full brow	>50%	43%
Flat midface	>50%	33%
Ptosis	>50%	4%
Large/prominent ears	>50%	65%
Wide nasal bridge	>50%	50%
Puffy cheeks	>50%	57%
Pointed chin	>50%	43%
Bulbous nose	>50%	59%
Widely spaced teeth/malocclusion	>25%	38%
Epicanthal folds	>25%	48%
Extremities		
Large and fleshy hands	>75%	48%
Dysplastic toenails	>75%	23%
2/3 rd toe syndactyly	>25%	44%
5 th finger clinodactyly	>25%	15%
Other features		
Decreased sensitivity to pain	>50%	69%
Hypohydrosis	>50%	17%
High tolerance to pain		69% (9/13)
Brain abnormalities at MRI		69% ^b
Cardiac anomalies	6% ^c	6,9%
Renal problems	>25% ^d	10%
Behavioural features		
Autism (DMS-IV)		13%
Behavioural disturbances		77% ^e