

Supplementary 2. Clinical presentation of current patients versus previously published cases (Parida et al.³⁾)

Variable	Reported Frequency (%) (Parida et al.)	Case 1	Case 2
Postnatal growth retardation	60%	Yes	No
Microcephaly	53%	No	No
Cerebellar hypoplasia (on MRI)	40%	Yes	NA
Dysmorphic facial features	60%	No	No
Hip dislocation (congenital)	6.6%	No	No
Hand deformity	53%	Yes	Yes
Foot Deformity	47%	Yes	Yes
Myopathic facies	40%	Yes	Yes
Sensory-neural deafness	73%	Yes	Yes
Exocrine pancreatic abnormality	40%	No	No
Hypothyroidism	20%	No	No
Diabetes mellitus	26.6%	Yes	No
Diabetic ketoacidosis	6.6%	No	No
Motor delay	93%	Yes	Yes
Intellectual disabilities	93%	Yes	Yes
Seizures	26.6%	Yes	Yes
Ataxia	66.6%	No	No
Hypoplasia of cerebellum	40%	No	No
Demyelinating peripheral neuropathy	47%	Yes	Yes

MRI, magnetic resonance imaging; NA, not available.