

Table 1. Summary of clinical, radiological, and biochemical findings in the affected individuals.

	Patient 1	Patient 2
Age	14 years	13 months
Gender	Male	Female
Height (SD)	149 cm (4 th centile)	74 cm (40 th centile)
Weight (SD)	45 kg (25 th centile)	7.25 kg (-2.83 SD)
Head circumference (SD)	55 cm (50 th centile)	41.5 cm (-3.2 SD)
Consanguinity	Yes	Yes
Abnormal fingernails and toenails	Yes	Yes
Triphalangeal thumb	No	Yes
Developmental delay or intellectual disability	Yes	Yes
Seizure	GTCS	None noted, but tonic posturing present
Deafness	No	No
MRI brain	Normal	Normal
Serum alkaline phosphatase level	295 IU/L (normal)	210 IU/L (normal)

MRI: magnetic resonance imaging; GTCS: generalised tonic-clonic seizure; NA: not available