

SUPPLEMENTARY TABLE S2. PHENOTYPE DETAILS OF AFFECTED INDIVIDUALS FROM FAMILY 2

<i>Phenotype</i>	<i>Family2.III.1 (Proband)</i>	<i>Family2.III.2 (Brother)</i>
Age	14	7
ID	Moderate	Moderate
Independent walking (years)	5	5
First words (years)	6	5
Speaking in sentences	Not attained	Not attained
Seizure	No	Myoclonic seizures
Head circumference	49 cm (3.7 SD below mean)	47.5 cm (3.6 SD below mean)
Microcephaly	Yes	Yes
Height	150 cm (2 SD below mean)	104 cm (3 SD below mean)
Anterior Hairline	Low	Low
Anterior cow-lick	Present	Absent
Eye brows	Straight	Straight
Synophrys	No	Yes
Nostril	Normal	Flaring
Mouth	Open/closed	Open
Ear position	Low set and posterior	Low set and posterior
Clinodactyly	Yes	Yes
Simian crease	No	Yes
Joints	—	Hyperextensible
Limbs	Long	—
Skin	Dry	Dry
Deep tendon reflexes	Brisk	Brisk
Gait	Normal	Normal
Co-ordination	Normal (no ataxia)	Normal (no ataxia)
Behavior	ADHD	ADHD (self injurious behavior)