

SUPPLEMENTARY TABLE S3. PHENOTYPE DETAILS OF AFFECTED INDIVIDUALS FROM FAMILY 3

<i>Phenotype</i>	<i>Family3.III.1 (Proband)</i>	<i>Family3.III.2 (twin brother)</i>
Age (years)	9	9
ID	Moderate	Borderline
IQ	42	72
Independent walking (months)	15	13
First words (years)	5	2
Talk in sentences	Not attained	3 years
Head circumference (cm)	52	51
Height (cm)	142	138
Anterior hairline	Low	Typical
Synophrys	Yes	Yes
High arched palate	Yes	Yes
Long/prominent ears	Yes	Yes
Hearing	Bilateral sensory-neural hearing impairment	Bilateral sensory-neural hearing impairment
Neck	Broad	Broad
Gynecomastia	Mild	Mild
Enlarged nipples	Yes	Yes
Abdomen	Distended	Distended
Knees	Mild knock knees	Mild knock knees
Behavior	Autism spectrum disorder with ADHD	No abnormality