

**Additional file 2: Table S2**

**Medical Features**

Participant	Mutation	Seizures	Hypotonia	Recurring infection	Constipation/ diarrhea	Gastroesophageal reflux disease	History or regression	Gait abnormalities	Brain MRI abnormalities	Sleep disturbance
1	G1527A		x					x	x	
2	2497delG	x	x	x	x	x	x	x	x	x
3	A5008T	x	x	x	x	x	x	x	x	x

**Dysmorphic features**

Participant	Mutation	Long eyelashes	Ear anomalies	Full lips	Epicanthal folds	Macrocephaly	Dolicocephaly	Wide nasal bridge	Hypertelorism	Short stature	2-3 Syndactyly of toes	5th Finger clinodactyly	Deep set eyes	Broad nasal bridge
1	G1527A	x	x	x	x	x		x	x					x
2	2497delG						x			x				
3	A5008T										x	x	x	