

Supplementary Table 3 Additional health and medical phenotypic features in individuals with pathogenic missense/loss-of-function variants disrupting *FOXP2*

Case	Vision impairment	Physical features	Mental health	MRI/CT	Feeding	Drooling	Other medical
1a	-	Upturned nose, mild retrognathia, mildly prominent ears, full lips	-	Incidental L anterior temporal arachnoid cyst	-	+	Tongue tie, Hx ear infections
1b	-	Prominent nose with fleshy nasal tip, mild retrognathia, mildly prominent ears, full lips	-	Hypoplasia of vermis	-	-	Tonsillectomy
1c	Glasses, myopia	Prominent nose with fleshy nasal tip, full lips	Anxiety, depression	Atrophic cerebellar vermis, mega cisterna magna, mild ventriculomegaly	-	-	-
1d	Glasses, hypermetropia	NA	Anxious behaviour	NA	-	-	Moderate, mixed hearing impairment
2	-	Mildly wide mouth, mildly short philtrum	Anxious behaviour	NA	+	+	-
3a	Glasses, squint	Mild periorbital fullness, rounded nasal tip, mild retrognathia, mildly thin upper lip	-	NA	-	-	Stage 2 chronic kidney disease, milk allergy
3b	Glasses, myopia	-	-	NA	-	-	-
4a	-	Synophrys, mildly full lips, toe/toe nail abnormalities, large feet	OCD, anxiety, depression	NA	+ Hx	-	-
4b	-	Mildly full lips, mildly hypoplastic alar nasae, toe/toe nail abnormalities, large feet	OCD, anxiety, depression	NA	+ Hx	-	-
4c	-	Mildly hypoplastic alar nasae, toe/toe nail abnormalities	Anxiety, depression	NA	-	-	-

5	Strabismus	Mild periorbital fullness, rounded nasal tip	-	NA	+	-	Submucous cleft palate
6	-	Round face	-	MRI NAD	-	-	Eczema, hypotonia
7a	-	Epicanthic folds, mildly up slanting palpebral fissures, prominent incisors, sandal gaps	-	NA	-	NA	-
7b	-	Epicanthic folds, mildly up slanting palpebral fissures, prominent incisors, sandal gaps	-	NA	-	NA	-
8a	-	-	Depression	NA	-	NA	-
8b	-	High nasal root, a pointed nasal tip, prominent incisors, overbite, tapering fingers	-	NA	-	NA	Recurrent middle ear infections in infancy
9	-	-	-	NA	-	NA	-
10	NA	Narrow palpebral fissures, high arched palate, mild finger pads.	-	NA	+	NA	Possible seizure at 10mo
11a	Extropia, strabismus	-	NA	NA	NA	NA	NA
11b	NA	-	NA	NA	NA	NA	NA
12a	-	Flat face, upturned nose, small mouth, clinodactyly	-	NA	-	-	Hypotonia
12b	Glasses, myopia	Overbite, flat affect, protruding ears	-	NA	-	-	-
13	-	Full lips, mild periorbital fullness, slightly short palpebral fissures, mild retrognathia	-	NA	+	+	Laryngomalacia, tracheomalacia, laryngeal cleft
14	-	Mildly anteverted ears	-	CT NDA	+	-	-

15	-	-	-	CT NDA	+	-	Mild, bilateral, conductive hearing impairment, eczema
16a	Glasses, myopia	Overbite	Anxiety, depression	NA	-	-	Petite mal seizure unspecified x1 at 10yrs
16b	-	Round face, slightly prominent eyes	-	NA	+	+	Eczema, allergies, chronic constipation, seasonal asthma
17	-	Epicanthic folds, flat philtrum, single palmar crease right hand, microcephaly	-	MRI periventricular gliosis & right frontal migration abnormality	+	+	Eczema

+ = feature present, - = feature absent, CT = computed tomography, Hx = history (previous), mo = months, MRI = magnetic resonance imaging, NA = not applicable, NA = not assessed, NAD = no abnormalities detected, yrs = years