

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 | Droling | 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 | - | Synophyrus, 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 | - | - | - |

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|------------|----------------------|--------------------------------------------------------------------------------------------|------------|---------|----|----|-------------------------------------------------|
| 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 | + | - | - |

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|------------|-----------------|--------------------------------------------------------------------------------|---------------------|-------------------------------------------------------------------|---|---|----------------------------------------------------------|
| 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