**Table S1. Summary of clinical features identified in the cohort of 26 patients**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Features identified** | | | | | **Frequency**  **(n = 26)** | **Proportion (%)** |
| **Short stature** | | | | | 21 | 80.8 |
| **Central Nervous System** | Agenesis of the Corpus Callosum | | | | 1 | 3.8 |
| **Eye** | Strabismus | | | | 4 | 15.4 |
| Myopia | | | | 3 | 11.5 |
| Nystagmus | | | | 2 | 7.7 |
| Astigmatism | | | | 1 | 3.8 |
| **Auditory** | Conductive hearing loss | | | | 3 | 11.5 |
| Sensorineural hearing loss | | | | 1 | 3.8 |
| **Oral & Dental** | High arched palate | | | | 11 | 42.3 |
| Dental malocclusion | | | | 11 | 42.3 |
| Dental caries | | | | 2 | 7.7 |
| Widely spaced teeth | | | | 1 | 3.8 |
| Overcrowded teeth | | | | 1 | 3.8 |
| **Chest** | pectus excavatum (PE) | | | | 6 | 23.1 |
| Pectus carinatum (PC) | | | | 4 | 15.4 |
| PC superiorly & PE inferiorly | | | | 1 | 15.4 |
| **Musculoskeletal (other than chest)** | Spine | | Scoliosis | | 5 | 19.2 |
| Lumbar hyperlordosis | | 1 | 3.8 |
| Limbs | | Cubitus valgus | | 2 | 7.7 |
| Talipes equinovarus | | 1 | 3.8 |
| High arched feet sole | | 1 | 3.8 |
| Joints | | Hyperlaxity | | 3 | 11.5 |
| Contracture | | 2 | 7.7 |
| Other | | 5th finger clinodactyly | | 1 | 3.8 |
| Overlapping 2-3 toes | | 1 | 3.8 |
| Fused ribs | | 1 | 3.8 |
| **Gastrointestinal** | Feeding  difficulties | | Poor suck & prolonged  feeding time | | 3 | 11.5 |
| Severe feeding issues with tube-feeding for > 3 weeks | | 3 | 11.5 |
| Very poor suck & slow feeding with recurrent vomiting | | 2 | 7.7 |
| Pyloric stenosis | | | | 1 | 3.8 |
| **Genitourinary** | Undescended testis | | | | 7/15 males | 46.7 |
| Duplex left collecting system | | | | 1 | 3.8 |
| Multicystic kidney | | | | 1 | 3.8 |
| **Dermatology** | Thick curly hair | | | | 15 | 57.7 |
| Skin | Cafe-au-lait spots | | | 9 | 34.6 |
| Pigmented naevi | | | 3 | 11.5 |
| Keratosis pilaris atrophicans faciei | | | 1 | 3.8 |
| Axillary freckling | | | 1 | 3.8 |
| Hypopigmented facial lesion | | | 1 | 3.8 |
| **Haematology & Oncology** | Easy bruising | | | | 14 | 53.8 |
| Abnormal PT & PTT | | | | 1 | 3.8 |
| Mild thrombocytopenia | | | | 1 | 3.8 |
| Desmoid cyst | | | | 1 | 3.8 |
| **Lymphatic** | Lymphedema | | | | 3 | 11.5 |
| **Neurology, Cognition&**  **Behaviour** | Learning difficulties | | | | 11 | 42.3 |
| Mild intellectual disability | | | | 7 | 26.9 |
| Severe intellectual disability | | | | 1 | 3.8 |
| Behavioural issues | | | Hyperactive | 5 | 19.2 |
| ADHD | 4 | 15.4 |
| Self-injury | 1 | 3.8 |