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

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| --- | --- | --- |
| **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** | Feedingdifficulties | 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 |