**Table S5**. Summary of clinical characteristics associated with *(de novo) SATB1* PTVs and (partial) gene deletions predicted to result in haploinsufficiency and PTVs in the last exon.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | Individuals with PTVs and (partial) gene deletions predicted to result in haploinsufficiency | | Individuals with PTVs in the last exon | |
|  | **%** | **Present / total assessed** | **%** | **Present / total assessed** |
| Neurologic |  |  |  |  |
| Intellectual disability | 86 | 6/7 | 67 | 2/3 |
| Normal | 14 | 1/7 | 33 | 1/3 |
| Borderline | 0 | 0/7 | 0 | 0/3 |
| Mild | 71 | 5/7 | 33 | 1/3 |
| Moderate | 14 | 1/7 | 0 | 0/3 |
| Severe | 0 | 0/7 | 0 | 0/3 |
| Profound | 0 | 0/7 | 0 | 0/3 |
| Unspecified | 0 | 0/7 | 33 | 1/3 |
| Developmental delay | 100 | 7/7 | 100 | 5/5 |
| Motor delay | 86 | 6/7 | 100 | 5/5 |
| Speech delay | 86 | 6/7 | 80 | 4/5 |
| Dysarthria | 14 | 1/7 | 0 | 0/4 |
| Epilepsy | 0 | 0/6 | 40 | 2/5 |
| EEG abnormalities | 0 | 0/4 | 67 | 2/3 |
| Hypotonia | 43 | 3/7 | 40 | 2/5 |
| Spasticity | 0 | 0/7 | 0 | 0/5 |
| Ataxia | 14 | 1/7 | 20 | 1/5 |
| Behavioral disturbances | 100 | 7/7 | 0 | 0/5 |
| Sleep disturbances | 50 | 3/6 | 0 | 0/5 |
| Abnormal brain imaging | 33 | 1/3 | 50 | 2/4 |
| Regression | 14 | 1/7 | 0 | 0/5 |
| Growth |  |  |  |  |
| Abnormalities during pregnancy | 33 | 2/6 | 20 | 1/5 |
| Abnormalities during delivery | 33 | 2/6 | 80 | 4/5 |
| Abnormal term of delivery | 0 | 0/5 | 20 | 1/5 |
| Preterm (<37 weeks) | 0 | 0/5 | 20 | 1/5 |
| Postterm (>42 weeks) | 0 | 0/5 | 0 | 0/5 |
| Abnormal weight at birth | 20 | 1/5 | 25 | 1/4 |
| Small for gestational age (<p10) | 20 | 1/5 | 0 | 0/4 |
| Large for gestational age (>p90) | 0 | 0/5 | 25 | 1/4 |
| Abnormal head circumference at birth | 25 | 1/4 | 0 | 0/2 |
| Microcephaly\* | 0 | 0/4 | 0 | 0/2 |
| Macrocephaly# | 25 | 1/4 | 0 | 0/2 |
| Abnormal height | 14 | 1/7 | 0 | 0/4 |
| Short stature\* | 0 | 0/7 | 0 | 0/4 |
| Tall stature# | 14 | 1/7 | 0 | 0/4 |
| Abnormal head circumference | 0 | 0/5 | 25 | 1/4 |
| Microcephaly\* | 0 | 0/5 | 25 | 1/4 |
| Macrocephaly# | 0 | 0/5 | 0 | 0/4 |
| Abnormal weight | 0 | 0/5 | 25 | 1/4 |
| Underweight\* | 0 | 0/5 | 25 | 1/4 |
| Overweight# | 0 | 0/5 | 0 | 0/4 |
| Other phenotypic features |  |  |  |  |
| Facial dysmorphisms | 67 | 4/6 | 60 | 3/5 |
| Dental/oral abnormalities | 50 | 3/6 | 60 | 3/5 |
| Drooling/dysphagia | 29 | 2/7 | 20 | 1/5 |
| Hearing abnormalities | 17 | 1/6 | 20 | 1/5 |
| Vision abnormalities | 67 | 4/6 | 80 | 4/5 |
| Cardiac abnormalities | 17 | 1/6 | 40 | 2/5 |
| Skeleton/limb abnormalities | 33 | 2/6 | 0 | 0/5 |
| Hypermobility of joints | 33 | 2/6 | 25 | 1/4 |
| Gastrointestinal abnormalities | 33 | 2/6 | 20 | 1/5 |
| Urogenital abnormalities | 0 | 0/6 | 0 | 0/5 |
| Endocrine/metabolic abnormalities | 0 | 0/6 | 0 | 0/5 |
| Immunological abnormalities | 17 | 1/6 | 50 | 1/2 |
| Skin/hair/nail abnormalities | 0 | 0/6 | 20 | 1/5 |
| Neoplasms in medical history | 0 | 0/6 | 0 | 0/5 |
| \* <p3 |  |  |  |  |
| # >p97 |  |  |  |  |