**Supplementary Table 1:** Clinical features of the patient in comparison with the reported prevalence of 11p15 loss of methylation

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | | Patient | #Frequency in SRS patients with 11p15 loss of methylation (%) | \*Frequency in Japanese SRS patients with 11p15 loss of methylation (%) [n=43] |
| Key features in the Netchine-Harbison clinical scoring system | | | | |
|  | Small for gestational age | + | 100 (n=35) | NA |
| Postnatal growth failure | + | 83.8 (n=173) | NA |
| Relative macrocephaly at birth | + | 99.1 (n=112) | 100.0 |
| Protruding forehead | + | 93.7 (n=126) | 83.8 |
| Body asymmetry | + | 77.4 (n=226) | 81.1 |
| Feeding difficulties | - | 71.7 (n=173) | 47.1 |
| Low body mass index | + | NA | NA |
| Additional features | | | | |
|  | Triangular face | + | 98.7 (n=74) | 97.7 |
| Fifth finger clinodactyly | + | 80.7 (n=176) | 78.4 |
| Micrognathia | + | 74.7 (n=79) | NA |
| Irregular or crowded teeth | + | 28.6 (n=105) | 46.2 |
| Low-set and/or posteriorly rotated ears | + | 50.0 (n=140) | 40.0 |
| Down-turned mouth | + | 57.0 (n=114) | NA |
| High pitched or squeaky voice | + | 39.0 (n=26) | NA |
| Scoliosis and/or kyphosis | - | 10.0 (n=97) | NA |
| Toe syndactyly | - | 41.8 (n=141) | NA |
| Male genital abnormalities | + | 44.4 (n=63) | NA |
| Speech delay | + | 31.7 (n=101) | 25.8 |
| Motor delay | + | 30.5 (n=141) | 53.1 |
| Hypoglycemia | - | 21.7 (n=69) | NA |
| Excessive sweating | + | 51.4 (n=70) | NA |

# Wakeling EL, et al. Nat Rev Endocrinol 2017;13:105

\* Fuke T, et al. PLoS One 2013;8:e60105