**Supplementary table 1: Frequency of clinical features in patients with SRS and SGA**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  | **Total SRS**  **% (total no. of patients)** | **11p15LOM**  **% (total no. of patients)** | **upd(7)mat**  **% (total no. of patients)** | **Clinical SRS**  **% (total no. of patients)** |  | **non-SRS SGAb**  **% (total no. of patients)** | **Reference** |
| **NH-CSS Factors:** |  |  |  |  |  |  |  |  |
| Birth weight/height ≤ -2SDS a |  | 91.7 (60) | 100 (35) | 72.7 (11) | 85.7 (12) |  |  | 1 |
| Relative macrocephaly |  | 85.7 (209) | 99.1 (112) | 85.2 (27) | 55.4 (56) |  | 15.8 (146) | 1-4 |
| Postnatal height ≤ -2SDS |  | 84.2 (317) | 83.8 (173) | 80.9 (47) | 86.8 (83) |  | 87.7 (146) | 1-6 |
| Protruding forehead |  | 88.1 (201) | 93.7 (126) | 100.0 (27) | 77.6 (76) |  | 17.1 (146) | 1-4, 6 |
| Feeding difficulties |  | 70.4 (307) | 71.7 (173) | 87.2 (47) | 54.8 (73) |  | 25.3 (146) | 1-6 |
| Asymmetry |  | 57.3 (473) | 77.4 (226) | 29 (62) | 39.5 (157) |  | 6.8 (266) | 1-5, 7, 8 |
| **Additional Characteristics:** |  |  |  |  |  |  |  |  |
| Triangular face |  | 93.9 (164) | 98.7 (74) | 50.0 (16) | 98.7 (74) |  | 22 (90) | 2, 3, 6 |
| Delayed closure of fontanelle |  | 42.6 (47) | 44.4 (36) | 36.0 (11) | NDA |  | NDA | 5, 6 |
| Low-set &/or posteriorly rotated ears |  | 49.3 (266) | 50 (140) | 68.8 (48) | 35.9 (78) |  | 23.5 (98) | 1-3, 5 |
| Downturned mouth |  | 47.7 (176) | 57.0 (114) | 25.7 (39) | 39.1 (23) |  | 12.3 (98) | 1, 3, 5, 6 |
| Irregular/crowded teeth |  | 36.9 (195) | 28.6 (105) | 38.9 (36) | 24.1 (54) |  | 4.0 (90) | 2, 3, 5, 6 |
| Micrognathia |  | 61.7 (115) | 74.7 (79) | 25.9 (27) | 56.0 (9) |  | 23.0 (90) | 3, 5, 6 |
| Low muscle mass |  | 56.3 (103) | 67.2 (61) | 47.4 (19) | 34.8 (23) |  | 11.2 (98) | 1, 3 |
| 5th finger clinodactyly |  | 74.6 (319) | 80.7 (176) | 56.3 (48) | 77.8 (81) |  | 19.2 (146) | 1-6 |
| Syndactyly of toes |  | 29.9 (264) | 41.8 (141) | 16.7 (48) | 16.0 (75) |  | 11.2 (98) | 1-3, 5 |
| Prominent heels |  | 44.3 (61) | 25.7 (35) | 100 (12) | 42.9 (14) |  | 37.5 (8) | 1 |
| Shoulder dimples |  | 65.6 (61) | 77.1 (35) | 66.7 (12) | 35.7 (14) |  | 25.0 (8) | 1 |
| Scoliosis &/or Kyphosis |  | 17.6 (227) | 10.0 (97) | 16.3 (43) | 24.4 (78) |  | NDA | 3, 5, 9 |
| Male genital abnormalities |  | 40.0 (85) | 44.4 (63) | 21.4 (14) | 37.5 (8) |  | 21.2 (52) | 1, 3, 5, 6 |
| Motor delay |  | 36.6 (254) | 30.5 (141) | 58.3 (36) | 41.3 (63) |  | 18.8 (138) | 2-6 |
| Speech delay |  | 39.7 (189) | 31.7 (101) | 63.9 (36) | 38.5 (52) |  | 19.0 (90) | 2, 3, 5 |
| Autistic spectrum disorder / PDD |  | 18.0 (61) | 5.7 (35) | 58.3 (12) | 14.3 (14) |  | 12.5 (8) | 1 |
| High pitched/ squeaky voice |  | 45.2 (42) | 39 (26) | 71.0 ( 7) | 44.0 (9) |  | 12.0 (90) | 3 |
| Excessive sweating |  | 53.8 (106) | 51.4 (70) | 70.4 (27) | 22.0 (9) |  | 2.0 (90) | 3, 5 |
| Hypoglycaemia |  | 22.3 (103) | 21.7 (69) | 29 (0) | NDA |  | 6.2 (48) | 4, 5 |
| Abbreviations: MPTH, mid-parental target height; ND, no data available; NH-CSS, Netchine-Harbison clinical scoring system; PDD, pervasive developmental disorder  a In the majority of studies, SGA was mandatory for clinical diagnosis; the only one that did not & used either length or weight was Azzi et al. 1. **b**A single subject from this group was not SGA in the Azzi et al. 1 study but patient specific data was not available so percentages could not be altered. | | | | | | | | |

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