**Table 1:** Clinical findings of children with pathogenic copy number variations

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| Clinical findings | Frequency/total number of patients evaluated (%) |
| History of preterm birth | 4/26 (15.4) |
| Birth weight |  |
| AGA | 20/26 (76.9) |
| SGA | 5/26 (19.2) |
| LGA | 1/26 (3.8) |
| Parental consanguinity  | 8/26 (30.8) |
| Positive first degree family history | 3/26 (11.5) |
| Global developmental delay | 15/26 (57.7) |
| Mild | 8 |
| Moderate to severe | 7 |
| Intellectual disability | 7 (26.9) |
| Mild | 5 |
| Moderate to severe | 2 |
| Autism spectrum disorder | 5 (19.2) |
| Head circumference |  |
| Microcephaly | 7/26 (26.9) |
| Macrocephaly | 2/26 (7.7) |
| Tonus  |  |
| Hypertonicity | 14/26 (53.8) |
| Hypotonicity | 2/26 (7.7) |
| Facial dysmorphism | 20/26 (76.9) |
| Hearing impairment | 2/26 (7.7) |
| Visual impairment | 3/26 (11.5) |
| Epilepsy (seizure control is defined in 8 patients) | 9/26 (34.6) |
| Seizure control with monotherapy | 6/8 |
| Seizure control with two antiepileptic drugs | 1/8 |
| Drug-resistant epilepsy | 1/8 |
| EEG findings | 6/14 (42.9) |
| Focal anomaly  | 3 |
| Generalized anomaly  | 3 |
| Abnormal cranial MRI | 6/20 (30) |
| Accompanying major anomaly | 6/20 (30) |