Kleefstra Syndrome

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Kleefstra syndrome (KS), previously referred to as 9q subtelomeric deletion syndrome (9qSTDS), is characterised by moderate to severe developmental delay/mental retardation, childhood hypotonia, and brachy-microcephaly (main clinical phenotype), midface hypoplasia, prognathism, lip and eyebrow shape anomalies. The true prevalence of KS is unknown, but it is estimated that it occurs with a frequency of 1/200.000 in cases with mental retardation. On literature search, approximately 110 patients have been reported so far. Genetic analysis should be planned and interdisciplinary monitoring should be provided in cases suspected to have KS. Key Words: Child, Genetic disorder, Kleefstra Syndrome, Dysmorphism.