Short stature-craniofacial anomalies-genital hypoplasia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Short stature-craniofacial anomalies-genital hypoplasia syndrome. ORPHA:2994

Short stature-craniofacial anomalies-genital hypoplasia syndrome is characterized by the association of short stature, craniofacial anomalies and genital hypoplasia. Intellectual deficit is also found in the majority of cases, sometimes together with pterygia. Less than 20 cases have been described so far. The mode of transmission is likely to be autosomal dominant with incomplete penetrance. The syndrome is caused by unbalanced reciprocal translocations of the distal parts of chromosomes 6q and 9p, leading to partial trisomy of the distal region of chromosome 6q and partial monosomy of the distal region of chromosome 9p.