Chondrodysplasia-disorder of sex development syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Chondrodysplasia-disorder of sex development syndrome. ORPHA:1422

Chondrodysplasia - disorder of sex development is an extremely rare disorder of sex development (see this term), reported in only two siblings (one terminated in pregnancy) to date, characterized by the clinical features of 46,XY complete gonadal dysgenesis (see this term; normal external female genitalia, lack of pubertal development, primary amenorrhea, and hypergonadotrophic hypogonadism) in association with severe dwarfism with generalized chondrodysplasia (bell-shaped thorax, micromelia, brachydactyly). Other reported features in the live sibling included eye anomalies (hypoplastic irides, myopia, coloboma of optic discs), dysmorphic features (deep-set eyes, upslanting palpebral fissures, puffy eyelids, large ears and mouth, mild prognathism), muscular hypoplasia, mild intellectual deficiency and severe microcephaly with cerebellar vermis hypoplasia. An autosomal recessive inheritance has been suggested.