XY type gonadal dysgenesis-associated anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. XY type gonadal dysgenesis-associated anomalies syndrome. ORPHA:1770

Gonadal dysgenesis with multiple anomalies is an association syndrome described only once in two sisters aged 1 1/2 and 8 1/2 years. They had a 46,XY karyotype, cleft lip and palate, preauricular pits, and a 'squashed down' appearance because of a short columella and small nares. Other anomalies included broad hands and feet, and a hypermuscular appearance. Cardiac, renal, musculoskeletal, and ectodermal anomalies were also present. Ectodermal defects included 'punched out scalp defects' and unusual positioning of hair whorls. They also had short stature, streak gonads, and mild developmental delay. The mode of inheritance is most likely autosomal recessive.