46,XX disorder of sex development-skeletal anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 46,XX disorder of sex development-skeletal anomalies syndrome. ORPHA:2975

46,XX disorder of sex development-skeletal anomalies syndrome is characterised by primary amenorrhoea, ambiguous external genitalia, and bone abnormalities (hypoplasia of the mandibular condyles, hypoplasia of the maxilla, ulnar dislocation of the radial heads, etc.). It has been described in two sisters born to consanguineous parents.