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X-linked skeletal dysplasia-intellectual disability syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked skeletal dysplasia-intellectual disability syndrome. ORPHA:1436*

Skeletal dysplasia-intellectual disability syndrome combines skeletal anomalies (short stature, ridging of the metopic suture, fusion of cervical vertebrae, thoracic hemivertebrae, scoliosis, sacral hypoplasia and short middle phalanges) and mild intellectual deficit. It has been described in four male cousins in three sibships. Glucose intolerance was present in three cases, and imperforated anus in one case. Carrier females had minor manifestations (fusion of cervical vertebrae and glucose intolerance). Transmission seems to be X-linked.