Orofaciodigital syndrome type 6

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Orofaciodigital syndrome type 6. ORPHA:2754

Joubert syndrome with orofaciodigital defect (or oral-facial-digital syndrome type 6, OFD6) is a very rare subtype of Joubert syndrome and related disorders (JSRD, see this term) characterized by the neurological features of JS associated with orofacial anomalies and often polydactyly.