Orofaciodigital syndrome type 1

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Orofaciodigital syndrome type 1. ORPHA:2750

Oral-facial-digital syndrome type 1 (OFD1) is a rare neurodevelopmental disorder in the ciliopathy group that is lethal in males and characterized by variable anomalies including external malformations (craniofacial and digital), and possible involvement of the central nervous system (CNS) and of viscera (kidneys, pancreas and ovaries) in females.