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Hypertelorism-preauricular sinus-punctual pits-deafness syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Hypertelorism-preauricular sinus-punctual pits-deafness syndrome. ORPHA:293958

Hypertelorism-preauricular sinus-punctual pits-deafness syndrome is a rare developmental defect during embryogenesis syndrome characterized by hypertelorism, bilateral preauricular sinus, bilateral punctal pits, lacrimal duct obstruction, hearing loss, abnormal palmar flexion creases and bilateral distal axial triradii. Shawl scrotum has also been reported.