Oculotrichoanal syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Oculotrichoanal syndrome. ORPHA:2717

Oculotrichoanal syndrome is a form of rare, multiple congenital anomalies/dysmorphic syndrome characterized by a combination of various nose, eye, gastrointestinal and genitourinary abnormalities. Clinical presentation is variable and often includes bifid and broad nasal tip, aberrant anterior hairline, coloboma, cryptophthalmos or unilateral anophthalmia, anal anomalies, and omphalocele. Intelligence and global development is normal.