Hallermann-Streiff syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hallermann-Streiff syndrome. ORPHA:2108

Hallermann-Streiff syndrome is a rare genetic syndrome characterized mainly by head and facial abnormalities such as bird-like facies (with beak-shaped nose and retrognathia), hypoplastic mandible, brachycephaly with frontal bossing, dental abnormalities (e.g. absence of teeth, natal teeth, supernumerary teeth, severe agenesis of permanent teeth, enamel hypoplasia) hypotrichosis, various ophthalmic disorders (e.g. congenital cataracts, bilateral microphthalmia, ptosis, nystagmus) and atrophy of skin (especially around the center of face and nose) as well as telangiectasia and proportionate short stature. Intellectual disability is reported in some cases.