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Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome. ORPHA:397973*

Intellectual disability-obesity-prognathism-eye and skin anomalies syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by mild to profound intellectual disability, delayed speech, obesity, ocular anomalies (blepharophimosis, blepharoptosis, hyperopic astigmatism, decreased visual acuity, strabismus, abducens nerve palsy, and/or accommodative esotropia), and dermal manifestations, such as chronic atopic dermatitis. Associated craniofacial dysmorphism includes macrocephaly, maxillary hypoplasia, mandibular prognathism, and crowding of teeth.