SRD5A3-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. SRD5A3-CDG. ORPHA:324737

SRD5A3-CDG is a rare, non X-linked congenital disorder of glycosylation due to steroid 5 alpha reductase type 3 deficiency characterized by a highly variable phenotype typically presenting with severe visual impairment, variable ocular anomalies (such as optic nerve hypoplasia/atrophy, iris and optic nerve coloboma, congenital cataract, glaucoma), intellectual disability, cerebellar abnormalities, nystagmus, hypotonia, ataxia, and/or ichthyosiform skin lesions. Other reported manifestations include retinitis pigmentosa, kyphosis, congenital heart defects, hypertrichosis and abnormal coagulation.