Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome. ORPHA:2743

Ophthalmoplegia-intellectual disability-lingua scrotalis syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by congenital, external, nuclear ophthalmoplegia, lingua scrotalis, progressive chorioretinal sclerosis and intellectual disability. Bilateral ptosis, bilateral facial weakness, Parinaud's syndrome, convergence paresis and myopia may be associated. There have been no further descriptions in the literature since 1975.