X-linked intellectual disability, Najm type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability, Najm type. ORPHA:163937

Najm type X-linked intellectual deficit is a rare cerebellar dysgenesis syndrome characterized by variable clinical manifestations ranging from mild intellectual deficit with or without congenital nystagmus, to severe cognitive impairment associated with cerebellar and pontine hypoplasia/atrophy and abnormalities of cortical development.