Cerebellar hypoplasia-tapetoretinal degeneration syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cerebellar hypoplasia-tapetoretinal degeneration syndrome. ORPHA:2246

Cerebellar hypoplasia-tapetoretinal degeneration syndrome is a rare syndrome with a cerebellar malformation as a major feature characterized by cerebellar hypoplasia, bilateral retinal pigmentary changes, intellectual disability that can range from mild to moderate and pronounced language development delay. It presents with early developmental delay, central and peripheral non-progressive visual impairment or asymptomatic retinal changes, hypotonia, non-progressive ataxia and nystagmus.