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X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome. ORPHA:1568*

X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (XDIBS), or Pettigrew syndrome is a central nervous system malformation characterized by severe intellectual deficit, early hypotonia with progression to spasticity and contractures, choreoathetosis, seizures, dysmorphic face (long face with prominent forehead), and brain imaging abnormalities such as Dandy-Walker malformation (see this term), and iron deposition.