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Focal epilepsy-intellectual disability-cerebro-cerebellar malformation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Focal epilepsy-intellectual disability-cerebro-cerebellar malformation. ORPHA:352587

Focal epilepsy-intellectual disability-cerebro-cerebellar malformation is a rare, genetic neurological disorder characterized by early infantile-onset of seizures, borderline to moderate intellectual disability, cerebellar features including dysarthria and ataxia and cerebellar atrophy and cortical thickening observed on MRI imaging. Seizures are typically focal (with prominent eye blinking, facial and limb jerking), precipitated by fever and often commence with an oral sensory aura (anesthetized tongue sensation). When not properly controlled by anti-epileptic medication, weekly frequency and persistence into adult life is observed.