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Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression. ORPHA:391316*

Infantile-onset mesial temporal lobe epilepsy with severe cognitive regression is a rare monogenic disease with infantile-onset pharmacoresistant focal seizures of mesial temporal lobe onset manifesting with unresponsiveness, hypertonia and automatisms and cognitive regression soon after seizure onset leading to severe intellectual disability with behavioral abnormalities.