Hyperekplexia-epilepsy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyperekplexia-epilepsy syndrome. ORPHA:163985

A rare, X-linked, syndromic intellectual disability disease characterized by neonatal hypertonia which evolves to hypotonia and an exaggerated startle response (to sudden visual, auditory or tactile stimuli), followed by the development of early-onset, frequently refractory, tonic or myoclonic seizures. Progressive epileptic encephalopathy, intellectual disability, and psychomotor development arrest, with subsecuent decline, may be additionally associated.