PEHO-like syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. PEHO-like syndrome. ORPHA:99807

PEHO-like syndrome is a rare, genetic neurological disease characterized by progressive encephalopathy, early-onset seizures with a hypsarrhythmic pattern, facial and limb edema, severe hypotonia, early arrest of psychomotor development and craniofacial dysmorphism (evolving microcephaly, narrow forehead, short nose, prominent auricles, open mouth, micrognathia), in the absence of neuro-ophthalmic or neuroradiologic findings. Poor visual responsiveness, growth failure and tapering fingers are also associated.