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Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome. ORPHA:411986*

Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome is a rare, syndromic intellectual disability syndrome characterized by cortical blindness, different types of seizures, intellectual disability with limited or absent speech, and dysmorphic facial features. Brain imaging typically shows mild pontine hypoplasia, hypoplasia of the corpus callosum and atrophy in the occipital region.