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Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome. ORPHA:369939*

Severe motor and intellectual disabilities-sensorineural deafness-dystonia syndrome is a rare genetic neurological disorder characterized by intrauterine growth retardation, failure to thrive, infantile onset of sensorineural deafness, severe global developmental delay or absent psychomotor development, paraplegia or quadriplegia with dystonia and pyramidal signs, microcephaly, ocular abnormalities (strabismus, optic atrophy), mildly dysmorphic features (deep-set eyes, prominent nasal bridge, micrognathia), seizures and abnormalities of brain morphology (hypomyelinating white matter changes, cerebral atrophy).