Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome.
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Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome is a rare, genetic, neurometabolic disorder characterized by severe, progressive microcephaly, severe to profound global development delay, intellectual disability, seizures (typically tonic and/or myoclonic and frequently intractable), hyperekplexia, and axial hypotonia with appendicular spasticity, as well as hyperreflexia, dyskinetic quadriplegia, and abnormal brain morphology (cerebral atrophy with variable additional features including ventriculomeglay, pons and/or cerebellar hypoplasia, simplified gyral pattern and delayed myelination). Cortical blindness, feeding difficulties and respiratory insufficiency may also be associated.