Open Peer Review on Qeios

ITPA-related encephalopathy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ITPA-related encephalopathy. ORPHA:457375

A rare, genetic, neurometabolic disease characterized by early onset encephalopathy with progressive microcephaly, severe global development delay, seizures, hypotonia, feeding difficulties, variable cardiac abnormalities, and cataracts. Brain MRI shows distinct pattern with high T2 signal and restricted diffusion in the posterior limb of the internal capsule in combination with delayed myelination and progressive cerebral atrophy. The disease is typically fatal.