Open Peer Review on Qeios

Zellweger syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Zellweger syndrome. ORPHA:912

Zellweger syndrome (ZS) is the most severe variant seen in the peroxisome biogenesis disorders, Zellweger syndrome spectrum (PBD-ZSS; see this term), characterized by neuronal migration defects in the brain, dysmorphic craniofacial features, profound hypotonia, neonatal seizures, and liver dysfunction.