Infantile osteopetrosis with neuroaxonal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Infantile osteopetrosis with neuroaxonal dysplasia. ORPHA:85179

This syndrome is characterized by osteopetrosis, agenesis of the corpus callosum, cerebral atrophy and a small hippocampus.