Mitochondrial membrane protein-associated neurodegeneration

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mitochondrial membrane protein-associated neurodegeneration. ORPHA:289560

Mitochondrial membrane protein-sssociated neurodegeneration (MPAN), also known as neurogeneration with brain iron accumulation (NBIA) due to C19orf12 mutations, is an autosomal recessive neurodegenerative disorder characterized by iron accumulation in specific regions of the brain, usually the basal ganglia, and associated with slowly progressive pyramidal (spasticity) and extrapyramidal (dystonia) signs, motor axonal neuropathy, optic atrophy, cognitive decline, and neuropsychiatric abnormalities.