Mitochondrial DNA depletion syndrome, hepatocerebrorenal form

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Mitochondrial DNA depletion syndrome, hepatocerebrorenal form. ORPHA:363534

#64258;exia, ataxia, sensory neuropathy, epilepsy, sensorineural hearing impairment, psychomotor regression, athetosis, nystagmus, and/or ophthalmoplegia. Patients typically present with recurrent vomiting, severe failure to thrive, feeding difficulties, and fasting hypoglycemia.