Progressive external ophthalmoplegia-myopathy-emaciation syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Progressive external ophthalmoplegia-myopathy-emaciation syndrome. ORPHA:352447

Progressive external ophthalmoplegia-myopathy-emaciation syndrome is a rare mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies characterized by progressive external ophthalmoplegia without diplopia, cerebellar atrophy, proximal skeletal muscle weakness with generalized muscle wasting, profound emaciation, respiratory failure, spinal deformity and facial muscle weakness (manifesting with ptosis, dysphonia, dysphagia and nasal speech). Intellectual disability, gastrointestinal symptoms (e.g. nausea, abdominal fullness, and loss of appetite), dilated cardiomyopathy and renal colic have also been reported.