DNA2-related mitochondrial DNA deletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. DNA2-related mitochondrial DNA deletion syndrome. ORPHA:352470

A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by either late-onset myopathy with progressive external ophthalmoplegia and muscular weakness (predominantly limb-girdle) or early-onset myopathy presenting with decreased fetal movements, congenital ptosis, progressive external ophthalmoplegia, hypotonia and, variably, joint contractures. Reduced content and multiple deletions of mitochondrial DNA is observed in muscle biopsy.