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# Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency. ORPHA:329314*

Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency is an extremely rare multiple mitochondrial DNA deletion syndrome with markedly decreased deoxyguanosine kinase (DGUOK) activity in skeletal muscle characterized by a highly variable phenotype. Clinical manifestations include progressive external ophthalmoplegia, mitochondrial myopathy, recurrent rhabdomyolysis, lower motor neuron disease, mild cognitive impairment, sensory axonal neuropathy, optic atrophy, ataxia, hypogonadism and/or parkinsonism.