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Mitochondrial DNA depletion syndrome, myopathic form

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Mitochondrial DNA depletion syndrome, myopathic form. ORPHA:254875

Myopathic mitochondrial DNA (mtDNA) depletion syndrome is one of the main forms of mtDNA depletion syndrome (see this term) that displays a broad phenotypic spectrum but that is most often characterized by hypotonia, proximal muscle weakness, facial and bulbar weakness and failure to thrive.