# DISEASE:
# Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria

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| **NAME:** | Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria |
| **DESCRIPTION:** | A rare mitochondrial DNA depletion syndrome characterized by neonatal or infantile onset of global developmental delay, hypotonia, failure to thrive, progressive neurologic decline, sensorineural deafness, and movement disorder. Seizures, external ophthalmoplegia, polyneuropathy, cardiomyopathy, and renal tubular dysfunction have also been reported. Brain imaging may show T2-weighted hyperintensities in the basal ganglia, and laboratory examination may reveal lactic acidosis and mild methylmalonic aciduria. |
| **ORPHACODE:** | 1933 |
| **SYNONYMS:** | Booth-Haworth-Dilling syndrome<br>Mitochondrial encephalomyopathy-aminoacidopathy syndrome<br>mtDNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | SUCLA2 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Cholestasis (gene panel)
- Mitochondrial disorders (gene panel)
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)

## Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Brussel VUB

## Related Analytes

- succinate-CoA ligase ADP-forming subunit beta

## Related Gene Panels

- mitochondrial disease, nuclear based (343 genes) - VUB