

**DISEASE:**  
**Multiple mitochondrial dysfunctions syndrome type 1**

<b>NAME:</b>	Multiple mitochondrial dysfunctions syndrome type 1
<b>DESCRIPTION:</b>	A rare mitochondrial disease characterized by failure to thrive, infantile encephalopathy, muscular hypotonia, global developmental delay and regression, pulmonary arterial hypertension, episodes of apnea and bradycardia, respiratory failure, hyperglycinemia, and lactic acidosis. Hypertrophic or dilated cardiomyopathy have also been reported. Brain imaging may show leukoencephalopathy involving variable regions. The disease is typically fatal in early infancy.
<b>ORPHACODE:</b>	401869
<b>SYNOMYS:</b>	MMDS1 NFU1 deficiency
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">ICD-10</a> <a href="#">OMIM</a>
<b>ANALYTE(S):</b>	<a href="#">NFU1</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

## RELATED CONTENT

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### Related Genetic Tests

- Mitochondrial disorders (gene panel)

### Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

### Related Analytes

- NFU1 iron-sulfur cluster scaffold

### Related Gene Panels

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

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Source URL: <http://gentest.healthdata.be/disease/2272>