

Morava et al., 2006 criteria for mitochondrial disorder†

I. Clinical signs and symptoms

(1 point/symptom, max 4 points)

I.A. Muscular presentation (max 2 points)

- Ophthalmoplegia (2 points)
- Facies myopathica
- Exercise intolerance
- Muscle weakness
- Rhabdomyolysis
- Abnormal EMG

I.B. CNS presentation (max 2 points)

- Developmental delay
- Loss of skills
- Stroke-like episodes
- Migraine
- Seizures
- Myoclonus
- Cortical blindness
- Pyramidal signs
- Extrapyramidal signs
- Brain stem involvement

I.C. Multisystem disease (max 3 points)

- Hematology
- GI tract
- Endocrine/growth
- Heart
- Kidney
- Vision
- Hearing
- Neuropathy
- Recurrent/familial

Please consult your physician for help with interpretation

II. Metabolic/imaging studies

(1 point/symptom, max 4 points)

- Elevated lactate (2 points)
- Elevated lactate/pyruvate ratio
- Elevated alanine (2 points)
- Elevated CSF lactate (2 points)
- Elevated CSF protein
- Elevated CSF alanine (2 points)
- Urinary tricarbon acid excretion (2 points)
- Ethylmalonic aciduria
- Stroke-like picture MRI
- Leigh syndrome/MRI (2 points)
- Elevated lactate/MRS

III. Morphology (muscle biopsy)

(1 point/symptom, max 4 points)

- Ragged red/blue fibers (4 points)
- Cox-negative fibers (4 points)
- Reduced COX staining (4 points)
- Reduced SDH staining
- SDH positive blood vessels (2 points)
- Abnormal mitochondrial/EM (2 points)

Score:

- 1: mitochondrial disorder unlikely
- 2 to 4: possible mitochondrial disorder
- 5 to 7: probable mitochondrial disorder
- 8 to 12: definite mitochondrial disorder

†Reference:

Morava, E., L. van den Heuvel, F. Hol, M.C. de Vries, M. Hogeveen, R.J. Rodenburg, et al. (2006). Mitochondrial disease criteria: diagnostic applications in children. *Neurology*, 67, 1823-6.