

Mitochondrial disease: effect

What is a mitochondrial disease?

- Mitochondrial pathologies are genetic diseases due to mutations in nuclear and/or mitochondrial DNA that code for proteins found in mitochondria
- Since the mitochondria are the powerhouses of cells, the organs with the highest energy requirements (brain, muscle, heart) are mainly affected, but any organ can be involved

How many people are affected?

- In the UK, it has been estimated that 1 in 5,000 people are affected by mitochondrial disease

Can they be treated?

- No cure is currently available, and the only treatments are aimed at relieving symptoms

Nervous system and brain:

- Seizures
- Tremors
- Migraines/headaches
- Movement disorders
- Developmental delay
- Strokes before the age of 40
- Poor balance/coordination
- Encephalopathy
- Deafness
- Dementia

Eyes:

- Drooping eyelids (ptosis)



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- Blindness
- Cataracts

Heart:

- Cardiomyopathy

Skeletal muscle:

- Muscle weakness
- Exercise intolerance
- Cramps
- Pain
- Fatigue
- Low muscle tone

Liver:

- Liver failure
- Hypoglycemia

Kidneys:

- Renal failure
- Fanconi syndrome
- Nephrotic syndrome (uncommon, except for infants with Q₁₀ deficiency)

Digestive tract:

- Acid reflux
- Vomiting
- Diarrhea
- Difficulty swallowing
- Chronic constipation

Systemic

- Failure to gain weight
- Short stature

Pancreas and other glands

- Diabetes
- Parathyroid failure

