Mitochondrial disease: Effect
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What is a mitochondrial disease?
Mitochondrial diseases are genetic diseases due to mutations in both nuclear and mitochondrial DNA. 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.

What are the symptoms?
Mitochondrial diseases are incredibly variable in their symptoms, clinical features, age of onset and outcome. Typical manifestations include myopathy, ophthalmoplegia, stroke, epilepsy, blindness and deafness.

How many people are affected and can they be treated?
In the UK, it has been estimated that 1 in 5000 people are affected by mitochondrial disease. 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 delays
- Deafness
- Dementia
- Strokes before the age of 40
- Poor balance/ coordination
- Problems with peripheral nerves
- Encephalopathy

Eyes:
- Drooping eyelids (ptosis)
- Inability to move eyes from side-to-side (external ophthalmoplegia)
- Blindness (retinitis pigmentosa)
- Cataracts

Heart:
- Cardiomyopathy (heart failure, conduction block)

Liver:
- Liver failure (uncommon, except in babies with mtDNA depletion)
- Hypoglycemia (low blood sugar)

Digestive tract:
- Difficulty swallowing
- Acid reflux
- Vomiting
- Chronic constipation
- Diarrhea
- Irritable bowel-like symptoms

Systemic:
- Failure to gain weight
- Short stature

Kidneys:
- Renal failure
- Fanconi syndrome (loss of essential metabolites in urine)
- Nephrotic syndrome (uncommon, except for infants with Q10 deficiency)

Skeletal muscle:
- Muscle weakness
- Exercise intolerance
- Cramps
- Pain
- Fatigue
- Low muscle tone
- Excretion of muscle protein in urine (myoglobinuria)

Pancreas and other glands:
- Diabetes
- Parathyroid failure (low calcium)


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