Many mitochondrial disorders, resulting from mutations in both nuclear and mitochondrial DNA, have been characterised. Here, we focus on some of the diseases caused by mutations to mitochondrial DNA.

### LHON (Leber's hereditary optic neuropathy)
- **Mutations:**
  - $3460G>A$ (complex I: ND1)
  - $11778G>A$ (complex I: ND4)
  - $14484T>C$ (complex I: ND6)
- **Affects:** Retinal ganglion cells
- **Symptoms:** Sudden, painless loss of central vision, initially in one eye; second eye affected after 2 – 3 months

### MELAS (Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes)
- **Mutations:**
  - $3243A>G$ (transfer RNA: leu)
  - $3271T>C$ (transfer RNA: leu)
- **Affects:** Brain and nervous system
- **Symptoms:** Stroke-like episodes in the brain, migraines, vomiting, seizures; muscle weakness, exercise intolerance

### MERFF (Myoclonic Epilepsy with Ragged Red Fibers)
- **Mutations:**
  - $8344A>G$ (transfer RNA: lys)
  - $8356T>C$ (transfer RNA: lys)
- **Affects:** Muscles and nervous system
- **Symptoms:** Myoclonus (muscle jerks), seizures, ataxia and muscle weakness; deafness and short stature

### NARP (Neuropathy, ataxia, and retinitis pigmentosa)
- **Mutation:** $8993T>G$ (ATP synthase: ATP6)
- **Affects:** Nervous system
- **Symptoms:** NARP causes retinitis pigmentosa (blindness), neuropathy and ataxia

Other mitochondrial diseases involve large deletions of the mitochondrial genome:
- Kearns-Sayre syndrome: myopathy, ophthalmoplegia, cardiomyopathy
- CPEO: paralysis of eye movement