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

**LHON (Leber’s hereditary optic neuropathy)**
- Mutations: 3460G>A (complex I: subunit ND1)
  11778G>A (complex I: subunit ND4)
  14484T>C (complex I: subunit ND6)
- Affects: Retinal ganglion cells
- Symptoms: Sudden, painless loss of central vision. The classic pattern is for one eye to suddenly lose central vision, then two – three months later the other eye also loses central vision. After this “acute phase,” severe visual loss in both eyes remain.

**MERFF (Myoclonic Epilepsy with Ragged Red Fibers)**
- Mutations: 8344A>G (transfer RNA: lysine)
  8356T>C (transfer RNA: lysine)
- Affects: Muscles and nervous system
- Symptoms: The most prominent symptoms are myoclonus (muscle jerks), seizures, ataxia and muscle weakness. The disease also can cause hearing impairment and short stature.

**NARP (Neuropathy, ataxia, and retinitis pigmentosa)**
- Mutation: 8993T>G (ATP synthase: subunit ATP6)
- Affects: Nervous system
- Symptoms: NARP causes neuropathy (a malfunction of the nerves that can lead to sensory impairment and muscle weakness), ataxia and retinitis pigmentosa (degeneration of the retina in the eye, with resulting loss of vision). It also can cause developmental delay, seizures and dementia.

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Other mitochondrial diseases involve large deletions of the mitochondrial genome:
- Kearns-Sayre syndrome: Progressive myopathy, ophthalmoplegia, cardiomyopathy
- CPEO: The gradual paralysis of eye movement
- Pearson syndrome: This syndrome causes severe anemia and malfunction of the pancreas

Images: en.wikipedia.org and www.lhon.org/lhon/LHON.html