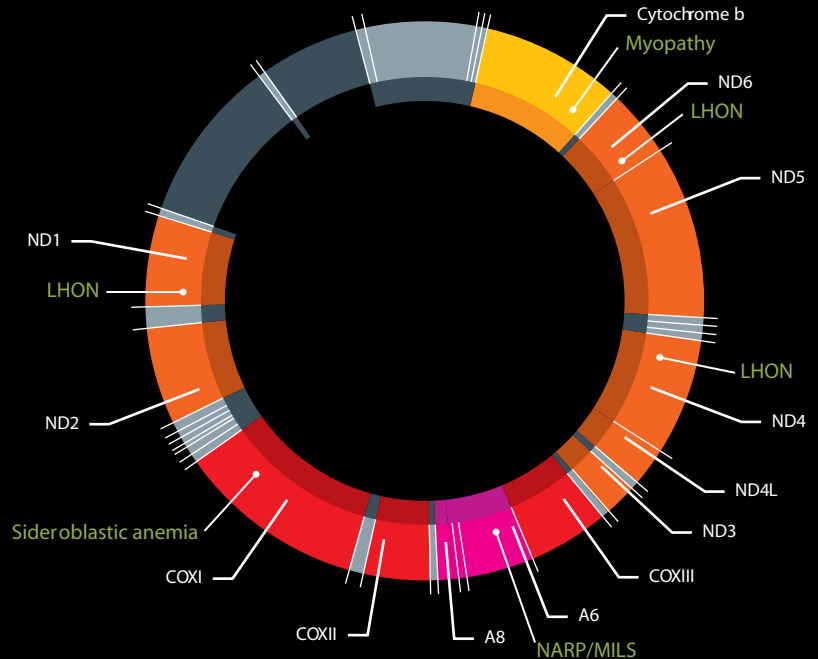


# When Mitochondria Mutate //

Sections of nuclear DNA dictate certain mitochondrial functions. When these sections mutate (in other words, when the DNA sequence within a gene is altered), mitochondria can malfunction, though the role of these malfunctions in various diseases—such as Parkinson’s and diabetes—remains murky. But mutations in the mitochondrion’s own 37 genes point more clearly to certain rare disorders. Here, dotted along the mitochondrion’s circular genome (its set of hereditary information), is a sampling of conditions relating to the 13 major genes that code for proteins that make up complexes. Each complex performs a specific step in the energy-producing chain.

- Complex I genes
- Complex III genes
- Complex IV genes
- Complex V genes
- Ribosomal RNA genes
- Transfer RNA genes



**LHON:** When inherited mutations cause complex I genes to malfunction, the result can be the death of retinal ganglion cells, whose axons compose the optic nerve. That can trigger the disease LHON (Leber’s hereditary optic neuropathy), which primarily strikes young men and results in the sudden loss of vision within just days or weeks.

**MYOPATHY:** Sporadic mutations (alterations in genes that accumulate during development or life and are not inherited) in the cytochrome b gene can cause a certain type of myopathy, or muscle disorder, which manifests as severe muscle weakness and the inability to exercise.

**SIDEROBLASTIC ANEMIA:** Sporadic mutations in the COX1 gene disrupt complex IV. This can lead to sideroblastic anemia, which results in fatigue, weakness, breathing difficulties and even liver damage and kidney failure. The mutation may impair the production of heme, the iron-rich part of blood cells that carries oxygen to the body’s tissues.

**NARP/MILS:** An inherited mutation in the A6 gene can lead to neuron death and brain atrophy. Diseases like NARP (neuropathy, ataxia and retinitis pigmentosa) and MILS (maternally inherited Leigh’s syndrome) can result, causing impairment in coordination, vision loss, dementia and seizures.