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MT-CO1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *MT-CO1 wt Allele*. NCI Thesaurus. Code C128942.

Human MT-CO1 wild-type allele is located within the circular mitochondrial (MT) chromosome and is approximately 1 kb in length. This allele, which encodes cytochrome c oxidase subunit 1, is involved in positive regulation of the mitochondrial electron transport chain. Allelic variation of the gene is associated with Leber hereditary optic neuropathy (LHON). Mutation of the gene is associated with mitochondrial complex IV deficiency (MT-C4D), recurrent mitochondrial myoglobinuria, and mitochondrially inherited nonsyndromic sensorineural deafness. Mutations may also be associated with colorectal cancer.