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OPHN1 wt Allele

National Cancer Institute

Source

National Cancer Institute. *OPHN1 wt Allele*. NCI Thesaurus. Code C127846.

Human OPHN1 wild-type allele is located in the vicinity of Xq12 and is approximately 392 kb in length. This allele, which encodes oligophrenin-1 protein, is involved in the regulation of small GTPase mediated signal transduction. Mutation of the gene is associated with X-linked mental retardation with cerebellar hypoplasia and distinctive facial dysmorphism.