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EDN3 wt Allele

National Cancer Institute

Source

National Cancer Institute. *EDN3 wt Allele*. NCI Thesaurus. Code C147999.

Human EDN3 wild-type allele is located in the vicinity of 20q13.32 and is approximately 26 kb in length. This allele, which encodes endothelin-3 protein, plays a role in vasoconstriction. Mutation of the gene is associated with congenital central hyperventilation syndrome, Waardenburg syndrome type 4B and increased susceptibility to Hirschsprung disease (aganglionic megacolon).