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EDNRB wt Allele

National Cancer Institute

Source

National Cancer Institute. *EDNRB wt Allele*. NCI Thesaurus. Code C105929.

Human EDNRB wild-type allele is located in the vicinity of 13q22 and is approximately 24 kb in length. This allele, which encodes endothelin receptor type B protein, plays a role in vasoconstriction, vasodilation, bronchoconstriction and cell proliferation. Mutations in this gene are associated with ABCD syndrome, Waardenburg syndrome, type 4A, and susceptibility to Hirschsprung disease 2.