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ATXN2 wt Allele

National Cancer Institute

Source

National Cancer Institute. *ATXN2 wt Allele*. NCI Thesaurus. Code C124927.

Human ATXN2 wild-type allele is located in the vicinity of 12q24.1 and is approximately 147 kb in length. This allele, which encodes ataxin-2 protein, is involved in the regulation of endocytosis of the epidermal growth factor receptor. Mutation of the gene is associated with spinocerebellar ataxia type 2, susceptibility to amyotrophic lateral sclerosis 13 and increased susceptibility for late-onset Parkinson disease.