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DES wt Allele

National Cancer Institute

Source

National Cancer Institute. *DES wt Allele*. NCI Thesaurus. Code C96449.

Human DES wild-type allele is located in the vicinity of 2q35 and is approximately 8 kb in length. This allele, which encodes desmin protein, plays a role in muscle cell contraction. Mutation of the gene is associated with myopathy myofibrillar desmin-related, cardiomyopathy dilated type 1I and neurogenic scapuloperoneal syndrome Kaeser type.