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Splice Site SNP

National Cancer Institute

Source

National Cancer Institute. *Splice Site SNP*. NCI Thesaurus. Code C45574.

An inherited single base substitution in a sequence of eukaryotic DNA located in either an acceptor (3' or downstream) or donor (5' or upstream) splice site of a gene. Functional single nucleotide polymorphisms in these intron-exon junctions can cause incorrect RNA splicing which, in turn, alters gene expression.