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JAK3 wt Allele

National Cancer Institute

Source

National Cancer Institute. *JAK3 wt Allele*. NCI Thesaurus. Code C51087.

Human JAK3 wild-type allele is located in the vicinity of 19p13.1 and is approximately 22 kb in length. This allele, which encodes tyrosine-protein kinase JAK3 protein, plays a role in signal transduction in the interleukin-2 and interleukin-4 signaling pathways. Mutations in the JAK3 gene which abolish the tyrosine-protein kinase activity of its protein product cause autosomal severe combined immunodeficiency disease.