PAX3 wt Allele

National Cancer Institute

Source

National Cancer Institute. PAX3 wt Allele. NCI Thesaurus. Code C52145.

Human PAX3 wild-type allele is located within 2q35-q37 and is approximately 99 kb in length. This allele, which encodes paired box gene Pax-3 protein, plays a role in transcriptional regulation that is critical for fetal development. Mutations in this gene are associated with Waardenburg syndrome, and craniofacial-deafness-hand syndrome. Also, mutations or translocations involving the gene are associated with alveolar rhabdomyosarcoma. The rhabdomyosarcoma-related PAX3 gene translocations include t(2;13)(q35;q14) with the FOXO1 gene and t(2;2)(q35;p23) with the NCOA1 gene.