FGFR1 wt Allele

National Cancer Institute

Source

National Cancer Institute. FGFR1 wt Allele. NCI Thesaurus. Code C51542.

Human FGFR1 wild-type allele is located within 8p11.2-p11.1 and is approximately 56 kb in length. This allele, which encodes basic fibroblast growth factor receptor 1 protein, is involved in the mediation of binding between both acidic and basic fibroblast growth factors and is involved in limb induction. Mutations in this gene are associated with several diseases, including Pfeiffer syndrome, Jackson-Weiss syndrome, Antley-Bixler syndrome, osteoglophonic dysplasia, and autosomal dominant Kallmann syndrome 2. Further, chromosomal aberrations involving the gene are associated with stem cell myeloproliferative disorder and stem cell leukemia lymphoma syndrome.