

FGFR4 gene

fibroblast growth factor receptor 4

Normal Function

The *FGFR4* gene provides instructions for making a protein called fibroblast growth factor receptor 4. This protein is part of a family of fibroblast growth factor receptors that share similar structures and functions. These receptor proteins play a role in important processes such as cell division, regulating cell growth and maturation, formation of blood vessels, wound healing, and embryo development.

The FGFR4 protein interacts with specific growth factors to conduct signals from the environment outside the cell to the nucleus. The nucleus responds to these signals by switching on or off appropriate genes that help the cell adjust to changes in the environment. In response, the cell might divide, move, or mature to take on specialized functions. Although specific functions of *FGFR4* remain unclear, studies indicate that the gene is involved in muscle development and the maturation of bone cells in the skull. The *FGFR4* gene may also play a role in the development and maintenance of specialized cells (called foveal cones) in the light-sensitive layer (the retina) at the back of the eye.

Health Conditions Related to Genetic Changes

Prostate cancer

MedlinePlus Genetics provides information about Prostate cancer

Cancers

A variation (polymorphism) in the *FGFR4* gene that causes a switch in amino acids (the building blocks of proteins) is associated with several types of cancer, such as those that occur in the breast, colon, head and neck, and prostate. In people with this polymorphism, glycine is replaced by arginine at position 388 in the protein's chain of amino acids. This variation is common and appears to occur in about 50 percent of humans. Although it produces no ill effects in healthy people, the mutation is associated with accelerated disease progression in certain cancers.

The abnormal activation and increased activity of the *FGFR4* gene are also implicated in the development of pituitary tumors and gastric, pancreatic, and ovarian cancers.

Other Names for This Gene

- CD334
- FGR4_HUMAN
- hydroxyaryl-protein kinase
- JTK2 Gene
- protein-tyrosine kinase
- TKF Gene
- tyrosylprotein kinase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FGFR4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2264\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2264[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FGFR4%5BTIAB%5D%29+OR+%28fibroblast+growth+factor+receptor+4%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- FIBROBLAST GROWTH FACTOR RECEPTOR 4; FGFR4 (<https://omim.org/entry/134935>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2264>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FGFR4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FGFR4[gene]))

References

- Bange J, Prechtel D, Cheburkin Y, Specht K, Harbeck N, Schmitt M, Knyazeva T, Muller S, Gartner S, Sures I, Wang H, Imyanitov E, Haring HU, Knayzev P, Iacobelli S, Hofler H, Ullrich A. Cancer progression and tumor cell motility are associated with the FGFR4 Arg(388) allele. Cancer Res. 2002 Feb 1;62(3):840-7. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11830541>)
- Cornish EE, Natoli RC, Hendrickson A, Provis JM. Differential distribution

offibroblast growth factor receptors (FGFRs) on foveal cones: FGFR-4 is an early marker of cone photoreceptors. *Mol Vis.* 2004 Jan 8;10:1-14. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14737068>)

- Eswarakumar VP, Lax I, Schlessinger J. Cellular signaling by fibroblast growth factor receptors. *Cytokine Growth Factor Rev.* 2005 Apr;16(2):139-49. doi:10.1016/j.cytogfr.2005.01.001. Epub 2005 Feb 1. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15863030>)
- Marics I, Padilla F, Guillemot JF, Scaal M, Marcelle C. FGFR4 signaling is a necessary step in limb muscle differentiation. *Development.* 2002 Oct;129(19):4559-69. doi: 10.1242/dev.129.19.4559. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12223412>)
- Streit S, Bange J, Fichtner A, Ihrler S, Issing W, Ullrich A. Involvement of the FGFR4 Arg388 allele in head and neck squamous cell carcinoma. *Int J Cancer.* 2004 Aug 20;111(2):213-7. doi: 10.1002/ijc.20204. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15197773>)
- Wang J, Stockton DW, Ittmann M. The fibroblast growth factor receptor-4 Arg388 allele is associated with prostate cancer initiation and progression. *Clin Cancer Res.* 2004 Sep 15;10(18 Pt 1):6169-78. doi: 10.1158/1078-0432.CCR-04-0408. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15448004>)

Genomic Location

The *FGFR4* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

Last updated June 1, 2005