

FZD4 Gene

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Frizzled class receptor 4

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1. Normal Function

The *FZD4* gene provides instructions for making a protein called frizzled-4. This protein is embedded in the outer membrane of many types of cells, where it is involved in transmitting chemical signals from outside the cell to the cell's nucleus. Specifically, frizzled-4 participates in the Wnt signaling pathway, a series of steps that affect the way cells and tissues develop. Wnt signaling is important for cell division (proliferation), attachment of cells to one another (adhesion), cell movement (migration), and many other cellular activities.

Studies suggest that, at the cell surface, the frizzled-4 protein interacts with a protein called norrin (produced from the *NDP* gene). The two proteins fit together like a key in a lock. Researchers suspect that when norrin attaches (binds) to frizzled-4, it initiates a multi-step process that regulates the activity of certain genes. During early development, signaling by norrin and frizzled-4 plays a critical role in the specialization of cells in the retina, which is the light-sensing tissue at the back of the eye. This signaling pathway is also involved in the establishment of a blood supply to the retina and the inner ear.

2. Health Conditions Related to Genetic Changes

2.1 Familial Exudative Vitreoretinopathy

More than 20 mutations in the *FZD4* gene have been identified in people with an eye disorder called familial exudative vitreoretinopathy. Some of these mutations change single protein building blocks (amino acids) in frizzled-4, while others insert or delete genetic material in the *FZD4* gene. Most *FZD4* mutations reduce the amount of frizzled-4 that is produced within cells. Other mutations are thought to result in the production of an unstable protein that cannot bind to norrin.

A reduction in the amount of frizzled-4 disrupts chemical signaling in the developing eye, which interferes with the formation of blood vessels at the edges of the retina. The resulting abnormal blood supply to this tissue leads to retinal damage and vision loss in some people with familial exudative vitreoretinopathy.

3. Other Names for This Gene

- CD344
- EVR1
- FEVR
- frizzled 4
- frizzled family receptor 4
- frizzled homolog 4 (Drosophila)
- Fz-4
- FZD4_HUMAN
- FZD4S

- FzE4
- GPCR
- MGC34390
- WNT receptor frizzled-4

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