FN1 Gene

Subjects: Genetics
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(This entry belongs to Entry Collection "MedlinePlus")

Definition

Fibronectin 1

1. Normal Function

The *FN1* gene provides instructions for making two types of the fibronectin-1 protein: soluble plasma fibronectin-1 and insoluble cellular fibronectin-1. Liver cells produce soluble plasma fibronectin-1 and release it into the bloodstream, where it is mainly involved in blood clotting and wound healing. Soluble plasma fibronectin-1 functions outside of cells (in the extracellular spaces), attaching (binding) to the surface of cells and binding to proteins, including other fibronectin-1 proteins. The attachment of these proteins form fibers that assist with tissue repair after an injury. Fibronectin-1 binding also helps with the continual formation of the extracellular matrix, which is an intricate lattice of proteins and other molecules that is made in the spaces between cells. This matrix provides structure and strength to tissues that support the body's organs. Many other cell types produce insoluble cellular fibronectin-1, which is released into the extracellular space and contributes to the creation of fibers and extracellular matrix. Both types of fibronectin-1 help individual cells expand (spread) and move (migrate) to cover more space and also influence cell shape and maturation (differentiation).

2. Health Conditions Related to Genetic Changes

2.1 Fibronectin Glomerulopathy

At least three mutations in the *FN1* gene have been found to cause fibronectin glomerulopathy, a progressive kidney disease that usually begins in adulthood and results in irreversible kidney failure (end-stage renal disease). *FN1* gene mutations account for about 40 percent of cases of fibronectin glomerulopathy. The *FN1* gene mutations change single protein building blocks (amino acids) in the fibronectin-1 protein. One mutation that occurs in multiple families replaces the amino acid tyrosine with the amino acid cysteine at position 973 in the fibronectin-1 protein (written as Tyr973Cys or Y973C). *FN1* gene mutations impair the protein's ability to bind to cells and proteins. The unbound fibronectin-1 protein, specifically soluble plasma fibronectin-1, is deposited in the glomeruli of the kidneys. These structures are clusters of tiny blood vessels in the kidneys that filter waste products from blood, which are then released in urine. Even though there is an abundance of fibronectin-1 in the glomeruli, the extracellular matrix that supports the blood vessels is weak because the altered fibronectin-1 cannot assist in the matrix's continual formation. Without a strong cellular support network, the glomeruli are less able to filter waste. As a result, products that normally are retained by the body, such as protein and blood, get released in the urine, and acids are not properly filtered from the blood. Over time, the kidneys' ability to filter waste decreases until the kidneys can no longer function, resulting in end-stage renal disease.

3. Other Names for This Gene

- CIG
- cold-insoluble globulin
- ED-B
- fibronectin

- FINC
- FINC_HUMAN
- FN
- FNZ
- LETS
- migration-stimulating factor
- MSF

References

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Keywords

genes