FREM1 Gene

Subjects: Genetics & Heredity

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FRAS1 related extracellular matrix 1

genes

1. Normal Function

The *FREM1* gene provides instructions for making a protein that is involved in the formation and organization of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues.

The FREM1 protein is one of a group of proteins, including proteins called FRAS1 and FREM2, that interact during embryonic development as components of basement membranes. Basement membranes help anchor layers of cells lining the surfaces and cavities of the body (epithelial cells) to other embryonic tissues, including those that give rise to connective tissues (such as skin and cartilage) and the kidneys.

2. Health Conditions Related to Genetic Changes

2.1 Manitoba Oculotrichoanal Syndrome

At least two *FREM1* gene mutations have been identified in people with Manitoba oculotrichoanal syndrome. This condition involves several characteristic physical features, particularly affecting the eyes (oculo-), hair (tricho-), and anus (-anal). The mutations delete genetic material from the *FREM1* gene or result in a premature stop signal that leads to an abnormally short FREM1 protein, and are believed to result in a nonfunctional protein.

Absence of functional FREM1 protein interferes with its role in embryonic basement membrane development and may also affect the location, stability, or function of the FRAS1 and FREM2 proteins. The features of Manitoba oculotrichoanal syndrome may result from the failure of neighboring embryonic tissues to fuse properly due to impairment of the basement membranes' anchoring function.

2.2 Coloboma

2.3 Congenital Anomalies of Kidney and Urinary Tract

2.4 Other Disorders

At least three *FREM1* gene mutations have been identified in people with a disorder called bifid nose, renal agenesis, and anorectal malformations syndrome, sometimes called BNAR. These mutations change single protein building blocks (amino acids) in the FREM1 protein or result in an abnormally shortened protein.

The mutations that cause BNAR likely disrupt the role of the FREM1 protein in the embryonic basement membranes of particular tissues, resulting in the cleft nasal cartilage (bifid nose), missing kidneys (renal agenesis), narrowed or misplaced anal opening (anorectal malformations), and other features characteristic of this disorder. Researchers suggest that BNAR and Manitoba oculotrichoanal syndrome, with their overlapping features, may be considered part of a single disorder spectrum.

3. Other Names for This Gene

- BNAR
- C9orf143
- C9orf145
- C9orf154
- extracellular matrix protein QBRICK
- FLJ25461
- FRAS1-related extracellular matrix protein 1
- FREM1_HUMAN
- TILRR

References

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