EFNB1 gene

ephrin B1

Normal Function

The *EFNB1* gene provides instructions for making a protein called ephrin B1. This protein spans the membrane that surrounds the cell. The portion outside the cell attaches (binds) to proteins called Eph receptor kinases on the surface of neighboring cells. Together, these proteins form Eph/ephrin complexes, which help cells stick to one another (cell adhesion) and communicate. Communication between the attached cells plays a critical role in the normal shaping (patterning) of many tissues and organs before birth. In the brain, Eph/ephrin complexes also play a part in the development of nerve cells (neurons) and in the ability of the connections between neurons (synapses) to change and adapt over time in response to experience (synaptic plasticity).

Health Conditions Related to Genetic Changes

Craniofrontonasal syndrome

More than 115 mutations in the *EFNB1* gene have been found to cause craniofrontonasal syndrome. This rare condition is characterized by the premature closure of certain bones of the skull (craniosynostosis) during development, which affects the shape of the head and face. Females with craniofrontonasal syndrome typically have more severe signs and symptoms than affected males, who often have one or two features of the condition.

Mutations in the *EFNB1* gene result in a shortage (deficiency) of ephrin B1 protein. Most of these mutations lead to an abnormally short version of the molecule that acts as the genetic blueprint used to make the ephrin B1 protein. The shortened molecules are quickly broken down before protein can be produced. A deficiency of ephrin B1 protein prevents the adhesion and communication between cells that aids in proper development , which disrupts normal patterning in tissues before birth. Abnormal development of the skull and other facial structures leads to the signs and symptoms of craniofrontonasal syndrome.

Other Names for This Gene

- EFL3
- EFNB1 gene

- ELK LIGAND
- Elk-L
- ELKL
- EPH-RELATED RECEPTOR TYROSINE KINASE LIGAND 2
- EPLG2
- LIGAND OF EPH-RELATED KINASE 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EFNB1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1947[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28EFNB1%5BTIAB%5D%29+OR+%28ephrin+B1%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- EPHRIN B1; EFNB1 (https://omim.org/entry/300035)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1947)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=EFNB1[gene])

References

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Genomic Location

The *EFNB1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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