FOXG1 gene

forkhead box G1

Normal Function

The *FOXG1* gene provides instructions for making a protein known as forkhead box G1. This protein is a transcription factor, which means it helps regulate the activity of other genes. Specifically, the forkhead box G1 protein acts as a transcriptional repressor, turning off (repressing) the activity of certain genes when they are not needed. Researchers believe that this protein plays an important role in brain development, particularly in a region of the embryonic brain known as the telencephalon. The telencephalon ultimately develops into several critical structures, including the the largest part of the brain (the cerebrum), which controls most voluntary activity, language, sensory perception, learning, and memory.

Health Conditions Related to Genetic Changes

FOXG1 syndrome

Changes involving the *FOXG1* gene cause *FOXG1* syndrome, a rare disorder characterized by impaired development and structural brain abnormalities. This condition was previously described as a congenital variant of Rett syndrome, which is a similar disorder of early development. However, doctors and researchers have identified some important differences between the two conditions, so now *FOXG1* syndrome is usually considered to be distinct from Rett syndrome.

At least 11 mutations within the *FOXG1* gene have been identified in people with *FOXG1* syndrome. The condition can also result from a deletion of genetic material from a region of the long (q) arm of chromosome 14 that includes the *FOXG1* gene and several neighboring genes. All of these genetic changes prevent the production of forkhead box G1 or impair the protein's function. A shortage of this protein disrupts normal brain development starting before birth, which appears to underlie the brain malformations and severe developmental problems characteristic of *FOXG1* syndrome.

Lennox-Gastaut syndrome

MedlinePlus Genetics provides information about Lennox-Gastaut syndrome

Other disorders

A few people have been found to have an extra copy (duplication) of the part of chromosome 14 that contains the *FOXG1* gene. These duplications are associated with recurrent seizures (epilepsy) starting in infancy, intellectual disability, and severe speech impairment. Duplications of the *FOXG1* gene have also been identified in several infants diagnosed with West syndrome, a condition characterized by epilepsy that begins in infancy, severe to profound intellectual disability, and related brain abnormalities. Although the mechanism is unclear, researchers believe that the extra genetic material leads to these developmental problems by altering early brain development.

Other Names for This Gene

- BF1
- BF2
- brain factor 1
- brain factor 2
- FHKL3
- FKH2
- forkhead box protein G1
- FOXG1_HUMAN
- FOXG1A
- FOXG1B
- FOXG1C
- HBF-1
- HBF-2
- HBF-3
- HBF-G2
- HBF2
- HFK1
- HFK2
- HFK3
- KHL2
- oncogene QIN
- QIN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of FOXG1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2290[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28FOXG1%5BTIAB%5D%29+OR+%28forkhead+box+G1%5BTIAB%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

- FORKHEAD BOX G1; FOXG1 (https://omim.org/entry/164874)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2290)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=FOXG1[gene])

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Genomic Location

The *FOXG1* gene is found on chromosome 14 (https://medlineplus.gov/genetics/chromosome/14/).

Last updated December 1, 2013