

## AFF2 gene

ALF transcription elongation factor 2

### Normal Function

The *AFF2* gene provides instructions for making a protein that is found in the nucleus of cells but whose function is not well understood. Some studies suggest that it acts as a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of other genes, although the identity of these genes is unknown.

Other studies show that the protein can attach to specific regions of messenger RNA (mRNA), which is a chemical cousin of DNA that serves as the genetic blueprint for protein production. It is thought that the *AFF2* protein helps control the process by which the mRNA blueprint is cut and rearranged to produce different versions of proteins (alternative splicing).

One region of the *AFF2* gene contains a particular DNA segment known as a CCG trinucleotide repeat, so called because this segment of three DNA building blocks (nucleotides) is repeated multiple times within the gene. In most people, the number of CCG repeats ranges from 4 to about 40.

### Health Conditions Related to Genetic Changes

#### Fragile XE syndrome

Mutations in the *AFF2* gene cause fragile XE syndrome, a condition characterized by mild intellectual disability and learning difficulties. Nearly all cases are caused by a mutation called a CCG trinucleotide repeat expansion, in which the CCG trinucleotide is abnormally repeated more than 200 times, which makes this region of the gene unstable. As a result, the *AFF2* gene is turned off (silenced), and no protein is produced from it. It is unclear how a shortage of this protein leads to problems with intellectual functioning.

Rarely, small deletions of genetic material from the *AFF2* gene are associated with fragile XE syndrome, although how these deletions affect the protein and lead to intellectual disability is unknown.

### Other Names for This Gene

- AF4/FMR2 family, member 2

- AFF2\_HUMAN
- FMR2
- FMR2P
- fragile X E mental retardation syndrome protein
- fragile X mental retardation 2 protein
- FRAXE
- MRX2
- OX19
- protein FMR-2

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of AFF2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2334\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2334[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28AFF2%5BTIAB%5D%29+OR+%28%28FMR2%5BTIAB%5D%29+OR+%28FMR2P%5BTIAB%5D%29+OR+%28fragile+X+mental+retardation+2+protein%5BTIAB%5D%29+OR+%28FRAXE%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+1080+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- ALF TRANSCRIPTION ELONGATION FACTOR 2; AFF2 (<https://omim.org/entry/300806>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2334>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=AFF2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AFF2[gene]))

## **References**

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## Genomic Location

The *AFF2* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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