

TFAP2A gene

transcription factor AP-2 alpha

Normal Function

The *TFAP2A* gene provides instructions for making a protein called transcription factor AP-2 alpha (AP-2 α). As its name suggests, this protein is a transcription factor, which means it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Transcription factor AP-2 α is one of a group of related proteins called AP-2 transcription factors. These proteins regulate genes that help control cell division and the self-destruction (apoptosis) of cells that are no longer needed.

Transcription factor AP-2 α is involved in development before birth. In particular, this protein is active in the neural crest, which is a group of cells in the early embryo that give rise to many tissues and organs. Among the embryonic structures formed from neural crest cells are the branchial arches, which develop into the bones and other tissues of the head and neck. The *TFAP2A* gene appears to be especially important for the development of tissues derived from the first and second branchial arches.

Health Conditions Related to Genetic Changes

Branchio-oculo-facial syndrome

Mutations in the *TFAP2A* gene cause a condition called branchio-oculo-facial syndrome, which is characterized by skin anomalies on the neck, malformations of the eyes and ears, and distinctive facial features. Most *TFAP2A* gene mutations involved in this condition change single protein building blocks (amino acids) in the transcription factor AP-2 α protein. These changes tend to occur in a region of the protein that enables it to bind to DNA. Although the effect of the amino acid changes on transcription factor AP-2 α is unknown, the protein's DNA binding function is likely impaired. Without this function, the protein cannot control the activity of genes during development. *TFAP2A* gene mutations disrupt the development of structures derived from the branchial arches, which results in the characteristic features of branchio-oculo-facial syndrome.

Anophthalmia/Microphthalmia

MedlinePlus Genetics provides information about Anophthalmia/Microphthalmia

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- activating enhancer-binding protein 2-alpha
- activator protein 2
- AP-2
- AP-2 transcription factor
- AP-2alpha
- AP2-alpha
- AP2A_HUMAN
- AP2TF
- BOFS
- TFAP2
- transcription factor AP-2 alpha (activating enhancer binding protein 2 alpha)
- transcription factor AP-2-alpha

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TFAP2A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7020\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7020[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TFAP2A%5BTIAB%5D%29+OR+%28transcription+factor+AP-2+alpha%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+1800+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- TRANSCRIPTION FACTOR AP2-ALPHA; TFAP2A (<https://omim.org/entry/107580>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7020>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TFAP2A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TFAP2A[gene]))

References

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Genomic Location

The *TFAP2A* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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