TFAP2A Gene

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Transcription factor AP-2 alpha: The TFAP2A gene provides instructions for making a protein called transcription factor AP-2 alpha (AP-2α).

Keywords: genes

1. Normal Function

The *TFAP2A* gene provides instructions for making a protein called transcription factor AP-2 alpha (AP-2α). 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-2α 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-2α 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.

2. Health Conditions Related to Genetic Changes

2.1. 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-2α 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-2α 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.

Coloboma

3. 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

References

1. de Crozé N, Maczkowiak F, Monsoro-Burq AH. Reiterative AP2a activity controls sequential steps in the neural crest gene regulatory network. Proc Natl Acad Sci U S A. 2011 Jan 4;108(1):155-60. doi: 10.1073/pnas.1010740107.
2. Developmental Biology (sixth edition, 2000): The Neural Crest
3. Hilger-Eversheim K, Moser M, Schorle H, Buettner R. Regulatory roles of AP-2transcription factors in vertebrate development, apoptosis and cell-cyclecontrol. Gene. 2000 Dec 30;260(1-2):1-12. Review.
4. Lin AE, Haldeman-Englert CR, Milunsky JM. Branchiooculofacial Syndrome. 2011May 31 [updated 2018 Mar 29]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE,Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK55063/
5. Milunsky JM, Maher TA, Zhao G, Roberts AE, Stalker HJ, Zori RT, Burch MN,Clemens M, Mulliken JB, Smith R, Lin AE. TFAP2A mutations result inbranchio-oculo-facial syndrome. Am J Hum Genet. 2008 May;82(5):1171-7. doi:10.1016/j.ajhg.2008.03.005. Erratum in: Am J Hum Genet. 2009 Feb;84(2):301..
6. Wang WD, Melville DB, Montero-Balaguer M, Hatzopoulos AK, Knapik EW. Tfap2aand Foxd3 regulate early steps in the development of the neural crest progenitor population. Dev Biol. 2011 Dec 1;360(1):173-85. doi: 10.1016/j.ydbio.2011.09.019.