TXNL4A gene

thioredoxin like 4A

Normal Function

The *TXNL4A* gene provides instructions for making one part (subunit) of a protein complex called the major spliceosome, which is the larger of two types of spliceosomes found in human cells. Spliceosomes help process messenger RNA (mRNA), which is a chemical cousin of DNA that serves as a genetic blueprint for making proteins. The spliceosomes recognize and then remove regions called introns to help produce mature mRNA molecules from immature mRNA molecules.

Health Conditions Related to Genetic Changes

Burn-McKeown syndrome

At least 10 *TXNL4A* gene mutations have been identified in people with Burn-McKeown syndrome, a disorder that is present from birth (congenital) and involves abnormalities of the nasal passages, characteristic facial features, hearing loss, heart abnormalities, and short stature. Most people with Burn-McKeown syndrome have a different genetic change in each of the two copies of the *TXNL4A* gene in each cell; this situation is called compound heterozygosity. In one copy of the gene, a *TXNL4A* gene mutation results in a protein with impaired function, or no protein at all is produced. The other copy of the *TXNL4A* gene has a deletion of a small amount of genetic material in an area near the *TXNL4A* gene called the promoter region, which controls the production of protein from that gene. This genetic change reduces protein production.

Research suggests that reduced quantities of the protein produced from the *TXNL4A* gene affect the assembly of the major spliceosome and change the production of a particular group of mRNA molecules. The details of these changes and their relationship to the specific signs and symptoms of Burn-McKeown syndrome are unknown. However, mutations in several genes involved in the spliceosome have been shown to cause other conditions with abnormalities affecting the head and face (craniofacial malformations), so craniofacial development is thought to be particularly sensitive to spliceosome problems.

Other Names for This Gene

- BMKS

- DIB1
- DIM1
- DIM1 protein homolog
- HsT161
- SNRNP15
- spliceosomal U5 snRNP-specific 15 kDa protein
- thioredoxin-like 4A
- thioredoxin-like U5 snRNP protein U5-15kD
- U5-15kD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TXNL4A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10907[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TXNL4A%5BTIAB%5D%29+OR+%28thioredoxin+like+4A%5BTIAB%5D%29%29+OR+%28%28BMKS%5BTIAB%5D%29+OR+%28DIB1%5BTIAB%5D%29+OR+%28DIM1+protein+homolog%5BTIAB%5D%29+OR+%28DIM1%5BTIAB%5D%29+OR+%28TXNL4%5BTIAB%5D%29+OR+%28U5-15kD%5BTIAB%5D%29+OR+%28spliceosomal+U5+snRNP-specific+15+kDa+protein%5BTIAB%5D%29+OR+%28thioredoxin-like+4A%5BTIAB%5D%29+OR+%28thioredoxin-like+4%5BTIAB%5D%29+OR+%28thioredoxin-like+U5+snRNP+protein+U5-15kD%5BTIAB%5D%29+OR+%28thioredoxin-like+protein+4A+isoform+1%5BTIAB%5D%29+OR+%28thioredoxin-like+protein+4A+isoform+2%5BTIAB%5D%29+OR+%28thioredoxin-like+protein+4A+isoform+3%5BTIAB%5D%29+OR+%28thioredoxin-like+protein+4A+isoform+4%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

- THIOREDOXIN-LIKE 4A; TXNL4A (https://omim.org/entry/611595)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/10907)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TXNL4A[gene])

References

- Lehalle D, Wieczorek D, Zechi-Ceide RM, Passos-Bueno MR, Lyonnet S, Amiel J, Gordon CT. A review of craniofacial disorders caused by spliceosomal defects.Clin Genet. 2015 Nov;88(5):405-15. doi: 10.1111/cge.12596. Epub 2015 May 1. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25865758)
- Simeoni F, Divita G. The Dim protein family: from structure to splicing. CellMol Life Sci. 2007 Aug;64(16):2079-89. doi: 10.1007/s00018-007-7043-9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17558560)
- Wieczorek D, Newman WG, Wieland T, Berulava T, Kaffe M, Falkenstein D, BeetzC, Graf E, Schwarzmayr T, Douzgou S, Clayton-Smith J, Daly SB, Williams SG, Bhaskar SS, Urquhart JE, Anderson B, O'Sullivan J, Boute O, Gundlach J, CzeschikJC, van Essen AJ, Hazan F, Park S, Hing A, Kuechler A, Lohmann DR, Ludwig KU,Mangold E, Steenpass L, Zeschnigk M, Lemke JR, Lourenco CM, Hehr U, Prott EC,Waldenberger M, Bohmer AC, Horsthemke B, O'Keefe RT, Meitinger T, Burn J, LudeckeHJ, Strom TM. Compound heterozygosity of low-frequency promoter deletions andrare loss-of-function mutations in TXNL4A causes Burn-McKeown syndrome. Am J HumGenet. 2014 Dec 4;95(6):698-707. doi: 10.1016/j.ajhg.2014.10.014. Epub 2014 Nov26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25434003) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4259969/)

Genomic Location

The *TXNL4A* gene is found on chromosome 18 (https://medlineplus.gov/genetics/chromosome/18/).

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