

EFTUD2 gene

elongation factor Tu GTP binding domain containing 2

Normal Function

The *EFTUD2* gene provides instructions for making one part (subunit) of two complexes called the major and minor spliceosomes. 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.

Health Conditions Related to Genetic Changes

Mandibulofacial dysostosis with microcephaly

More than 50 *EFTUD2* gene mutations have been identified in people who have mandibulofacial dysostosis with microcephaly (MFDM). This disorder causes malformations of the head and face, intellectual disability, and abnormalities affecting other areas of the body. These abnormalities include esophageal atresia, which is a blockage of the esophagus, and tracheoesophageal fistula, which is an abnormal connection between the esophagus and the trachea that allows fluids from the esophagus to get into the airways and interfere with breathing.

The *EFTUD2* gene mutations that cause MFDM result in the production of little or no functional enzyme from one copy of the gene in each cell. A shortage of this enzyme likely impairs mRNA processing. The relationship between these mutations and the specific symptoms of MFDM is not well understood.

Other Names for This Gene

- 116 kDa U5 small nuclear ribonucleoprotein component
- elongation factor Tu GTP-binding domain-containing protein 2
- hSNU114
- MFDGA
- MFDM
- SNRNP116
- Snrp116

- Snu114
- SNU114 homolog
- U5 snRNP-specific protein, 116 kDa
- U5-116KD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EFTUD2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9343\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9343[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28EFTUD2%5BTIAB%5D%29+OR+%28%28MFDG%5BTIAB%5D%29+OR+%28MFDGA%5BTIAB%5D%29+OR+%28Snu114%5BTIAB%5D%29+OR+%28Snrp116%5BTIAB%5D%29+OR+%28U5-116KD%5BTIAB%5D%29+OR+%28hSNU114%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%29>)

Catalog of Genes and Diseases from OMIM

- ELONGATION FACTOR Tu GTP-BINDING DOMAIN-CONTAINING 2; EFTUD2 (<https://omim.org/entry/603892>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9343>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EFTUD2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EFTUD2[gene]))

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Genomic Location

The *EFTUD2* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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