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.
Chromosomal Location

Cytogenetic Location: 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31

Molecular Location: base pairs 44,850,287 to 44,899,625 on chromosome 17 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

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
• U5-116KD
• U5 snRNP-specific protein, 116 kDa

Additional Information & Resources

Educational Resources


Clinical Information from GeneReviews

• Mandibulofacial Dysostosis with Microcephaly https://www.ncbi.nlm.nih.gov/books/NBK214367
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28EFTUD2%5BTIAB%5D%29+OR+%28%28MFDM%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

Catalog of Genes and Diseases from OMIM

- ELONGATION FACTOR Tu GTP-BINDING DOMAIN-CONTAINING 2
  http://omim.org/entry/603892

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EFTUD2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EFTUD2%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9343
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q15029

Sources for This Summary

- OMIM: ELONGATION FACTOR Tu GTP-BINDING DOMAIN-CONTAINING 2
  http://omim.org/entry/603892
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