



## EMD gene

emerin

### Normal Function

The *EMD* gene provides instructions for making a protein called emerin. Although this protein is produced in many tissues, it appears to be particularly important for the normal function of muscles used for movement (skeletal muscles) and the heart (cardiac muscle).

Within cells, emerin is a component of the nuclear envelope. The nuclear envelope is a structure that surrounds the nucleus, acting as a barrier between the nucleus and the surrounding fluid (cytoplasm) inside the cell. The nuclear envelope has several functions, including regulating the movement of molecules into and out of the nucleus.

Emerin interacts with several other proteins on the inner surface of the nuclear envelope. Together, these proteins are involved in regulating the activity of certain genes, controlling cell division and chemical signaling, and maintaining the structure and stability of the nucleus. Emerin and related proteins also play a role in assembling the nucleus during the process of cell division.

### Health Conditions Related to Genetic Changes

#### Emery-Dreifuss muscular dystrophy

More than 100 mutations in the *EMD* gene have been reported in people with Emery-Dreifuss muscular dystrophy. This condition affects skeletal and cardiac muscle, causing joint deformities called contractures, which restrict the movement of certain joints; muscle weakness and wasting that worsen over time; and heart problems, including an increased risk of sudden death.

Almost all of the *EMD* gene mutations prevent cells from producing any emerin protein. Researchers have not determined how a lack of this protein leads to the skeletal and cardiac muscle abnormalities characteristic of Emery-Dreifuss muscular dystrophy. Studies suggest, however, that an absence of emerin could disrupt the functions of other proteins in the nuclear envelope. These changes may alter the activity of certain genes or weaken the structure of the nucleus, making cells more fragile.

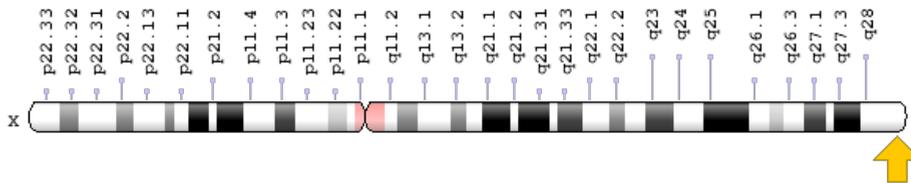
In rare cases, Emery-Dreifuss muscular dystrophy results from *EMD* mutations that change a single building block (amino acid) in the emerin protein. These mutations lead to the production of an abnormal version of emerin that is unable to interact with other proteins or cannot be correctly inserted into the nuclear envelope. This type of

mutation may be responsible for some cases of Emery-Dreifuss muscular dystrophy with unusually mild signs and symptoms.

### Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 154,379,236 to 154,381,523 on the X chromosome (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

### Other Names for This Gene

- EMD\_HUMAN
- emerin (Emery-Dreifuss muscular dystrophy)
- STA

### Additional Information & Resources

#### Educational Resources

- Madame Curie Bioscience Database: Lamins and Emerin in Muscular Dystrophy: The Nuclear Envelope Connection  
<https://www.ncbi.nlm.nih.gov/books/NBK6513/>
- Molecular Biology of the Cell (fourth edition, 2002): The Transport of Molecules between the Nucleus and the Cytosol  
<https://www.ncbi.nlm.nih.gov/books/NBK26932/>

#### Clinical Information from GeneReviews

- Emery-Dreifuss Muscular Dystrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK1436>

### Scientific Articles on PubMed

- PubMed  
[https://www.ncbi.nlm.nih.gov/pubmed?term=%28emerin%5BTIAB%5D%29+OR+%28%28EMD%5BTIAB%5D%29+AND+%28mutation\\*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D](https://www.ncbi.nlm.nih.gov/pubmed?term=%28emerin%5BTIAB%5D%29+OR+%28%28EMD%5BTIAB%5D%29+AND+%28mutation*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

### Catalog of Genes and Diseases from OMIM

- EMERIN  
<http://omim.org/entry/300384>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_EMD.html](http://atlasgeneticsoncology.org/Genes/GC_EMD.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=EMD%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:3331](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:3331)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:2010>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2010>
- UMD-EMD Mutations Database  
<http://www.umd.be/EMD/>
- UniProt  
<https://www.uniprot.org/uniprot/P50402>

### **Sources for This Summary**

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<https://ghr.nlm.nih.gov/gene/EMD>

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