

## MATR3 gene

matrin 3

### Normal Function

The *MATR3* gene provides instructions for making a protein called matrin 3, which is found in the nucleus of the cell as part of the nuclear matrix. The nuclear matrix is a network of proteins that provides structural support for the nucleus and aids in several important nuclear functions.

The function of the matrin 3 protein is unknown. This protein can attach to (bind) RNA, which is a chemical cousin of DNA. Some studies indicate that matrin 3 binds and stabilizes a type of RNA called messenger RNA (mRNA), which provides the genetic blueprint for proteins. Matrin 3 may also bind certain abnormal RNAs that could lead to nonfunctional or harmful proteins, thereby blocking the formation of such proteins. Other studies suggest that the matrin 3 protein may be involved in cell survival.

### Health Conditions Related to Genetic Changes

#### Distal myopathy 2

At least one mutation in the *MATR3* gene has been identified in people with distal myopathy 2, a condition characterized by muscle and vocal cord weakness. The *MATR3* gene mutation associated with distal myopathy 2 changes a single protein building block (amino acid) in the matrin 3 protein. This mutation, known as Ser85Cys (or S85C), replaces the amino acid serine with the amino acid cysteine at position 85 of the protein.

The effect of the S85C mutation on the function of the matrin 3 protein is unknown, although one study indicates that the mutation may change the location of the protein in the nucleus. Researchers are working to determine how this gene mutation leads to the signs and symptoms of distal myopathy 2.

#### Amyotrophic lateral sclerosis

MedlinePlus Genetics provides information about Amyotrophic lateral sclerosis

## Other Names for This Gene

- MATR3\_HUMAN
- matrin-3
- matrin-3 isoform a
- matrin-3 isoform b
- MGC9105
- MPD2

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MATR3 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9782\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9782[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MATR3%5BTIAB%5D%29+OR+%28matrin+3%5BTIAB%5D%29%29+OR+%28%28matrin-3%5BTIAB%5D%29+OR+%28MPD2%5BTIAB%5D%29+OR+%28VCPDM%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

- MATRIN 3; MATR3 (<https://omim.org/entry/164015>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9782>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MATR3\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MATR3[gene]))

## References

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## Genomic Location

The *MATR3* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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