

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

- Giordano G, Sanchez-Perez AM, Montoliu C, Berezney R, Malyavantham K, Costa LG, Calvete JJ, Felipe V. Activation of NMDA receptors induces protein kinase A-mediated phosphorylation and degradation of matrin 3. Blocking these effects prevents NMDA-induced neuronal death. J Neurochem. 2005 Aug;94(3):808-18. doi:10.1111/j.1471-4159.2005.03235.x. Epub 2005 Jul 5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16000164>)
- Przygodzka P, Boncela J, Cierniewski CS. Matrin 3 as a key regulator of endothelial

cell survival. *Exp Cell Res*. 2011 Apr 1;317(6):802-11. doi:10.1016/j.yexcr.2010.12.009. Epub 2010 Dec 21. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21182838>)

- Salton M, Elkon R, Borodina T, Davydov A, Yaspo ML, Halperin E, Shiloh Y. Matrin 3 binds and stabilizes mRNA. *PLoS One*. 2011;6(8):e23882. doi:10.1371/journal.pone.0023882. Epub 2011 Aug 17. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/21858232>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3157474/>)
- Salton M, Lerenthal Y, Wang SY, Chen DJ, Shiloh Y. Involvement of Matrin 3 and SFPQ/NONO in the DNA damage response. *Cell Cycle*. 2010 Apr 15;9(8):1568-76. doi:10.4161/cc.9.8.11298. Epub 2010 Apr 15. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20421735>)
- Zhang Z, Carmichael GG. The fate of dsRNA in the nucleus: ap54(nrb)-containing complex mediates the nuclear retention of promiscuously A-to-I edited RNAs. *Cell*. 2001 Aug 24;106(4):465-75. doi:10.1016/s0092-8674(01)00466-4. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11525732>)

Genomic Location

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

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