

MASP1 gene

MBL associated serine protease 1

Normal Function

The *MASP1* gene provides instructions for making proteins that are involved in a series of steps called the lectin complement pathway. This pathway is thought to help direct the movement (migration) of cells during development before birth to form the organs and systems of the body. It appears to be particularly important in directing the migration of neural crest cells. These cells give rise to various tissues including many tissues in the face and skull, the glands that produce hormones (endocrine glands), and portions of the nervous system. After birth, the lectin complement pathway is involved in the immune system.

Proteins called MASP-1, MASP-3, and MAp44 can be produced from the *MASP1* gene, depending on how the gene's instructions are pieced together. These proteins differ at one end of their structure. The MASP-1 and MASP-3 proteins have different versions of regions called serine protease domains, while the MAp44 protein has no serine protease domain. Researchers are studying whether these proteins play different roles in the lectin complement pathway.

Health Conditions Related to Genetic Changes

3MC syndrome

At least 11 *MASP1* gene mutations have been identified in people with 3MC syndrome, a disorder characterized by unusual facial features and a variety of problems affecting other tissues and organs of the body. Because all of the *MASP1* gene mutations that cause 3MC syndrome affect the MASP-3 protein, alterations in this protein's function are thought to account for the signs and symptoms of 3MC syndrome. Because parts of the three protein versions are the same, some of the *MASP1* gene mutations affect the other protein versions in addition to affecting MASP-3.

The protein changes result in faulty control of cell migration in early development, leading to the various abnormalities that occur in this disorder. Researchers suggest that similar pathways in the immune system can compensate for problems in the lectin complement pathway, which explains why immune system abnormalities are not part of 3MC syndrome.

Other Names for This Gene

- 3MC1
- complement factor MASP-3
- complement-activating component of Ra-reactive factor
- CRARF
- CRARF1
- mannan-binding lectin serine peptidase 1 (C4/C2 activating component of Ra-reactive factor)
- mannanose-binding lectin-associated serine protease 1
- mannanose-binding protein-associated serine protease
- MAP1
- MAp44
- MASP
- MASP3
- PRSS5
- Ra-reactive factor serine protease p100
- RaRF
- serine protease 5

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MASP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5648\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5648[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MASP1%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+peptidase+1%5BTIAB%5D%29%29+OR+%28%283MC1%5BTIAB%5D%29+OR+%28CRARF%5BTIAB%5D%29+OR+%28MAP1%5BTIAB%5D%29+OR+%28MASP3%5BTIAB%5D%29+OR+%28MASP%5BTIAB%5D%29+OR+%28MAp44%5BTIAB%5D%29+OR+%28Ra-reactive+factor+serine+protease+p100%5BTIAB%5D%29+OR+%28RaRF%5BTIAB%5D%29+OR+%28complement+factor+MASP-3%5BTIAB%5D%29+OR+%28complement-activating+component+of+Ra-reactive+factor%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+protease+1+isoform+1+precursor%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+protease+1+isoform+2+precursor%5BTIAB%5D%29+OR+%28mannan-binding+lectin+serine+protease+1+isoform+3+precursor%5BTIAB%5D%29+OR+%28mannose-binding+lectin-associated+serine+protease+1%5BTIAB%5D%29+OR+%28mannose-binding+protein-associated+serine+protease%5BTIAB%5D%29+OR+%28serine+protease+5%5BTIAB%5D%29%29+AND+%28%28Genes%5B>)

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Catalog of Genes and Diseases from OMIM

- MANNAN-BINDING LECTIN SERINE PROTEASE 1; MASP1 (<https://omim.org/entry/600521>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5648>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MASP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MASP1[gene]))

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Genomic Location

The *MASP1* gene is found on chromosome 3 (<https://medlineplus.gov/genetics/chromosome/3/>).

Last updated July 1, 2018