

## **MSTN gene**

myostatin

### **Normal Function**

The *MSTN* gene provides instructions for making a protein called myostatin. This protein is part of the transforming growth factor beta (TGF $\beta$ ) superfamily, which is a group of proteins that help control the growth and development of tissues throughout the body. Myostatin is found almost exclusively in muscles used for movement (skeletal muscles), where it is active both before and after birth. This protein normally limits muscle growth, ensuring that muscles do not grow too large. Myostatin has been studied extensively in mice, cows, and other animals, and it appears to have a similar function in humans.

Researchers are studying myostatin as a potential treatment for various muscular dystrophies that cause muscle weakness and wasting (atrophy).

### **Health Conditions Related to Genetic Changes**

#### Myostatin-related muscle hypertrophy

At least one variant (also known as a mutation) in the *MSTN* gene has been found to cause myostatin-related muscle hypertrophy, a rare condition characterized by increased muscle mass and strength. The variant, which is written as IVS1+5G>A, disrupts the way the gene's instructions are used to make myostatin. As a result, cells produce little or no functional myostatin. A loss of this protein in muscle cells leads to an overgrowth of muscle tissue. It does not appear to cause any other medical problems in affected individuals.

### **Other Names for This Gene**

- GDF-8
- GDF8
- GDF8\_HUMAN
- growth differentiation factor 8

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MSTN ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2660\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2660[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MSTN%5BTIAB%5D%29+OR+%28myostatin%5BTIAB%5D%29%29+OR+%28%28GDF-8%5BTIAB%5D%29+OR+%28GDF8%5BTIAB%5D%29+OR+%28growth+differentiation+factor+8%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+1800+days%22%5Bdp%5D%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- MYOSTATIN; MSTN (<https://omim.org/entry/601788>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2660>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MSTN\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MSTN[gene]))

## References

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## **Genomic Location**

The *MSTN* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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