

Myostatin-related muscle hypertrophy

Description

Myostatin-related muscle hypertrophy is a rare condition characterized by reduced body fat and increased muscle size. Affected individuals have up to twice the usual amount of muscle mass in their bodies. They also tend to have increased muscle strength. Myostatin-related muscle hypertrophy is not known to cause any medical problems, and affected individuals are intellectually normal.

Frequency

The prevalence of this condition is unknown.

Causes

Variants (also known as mutations) in the *MSTN* gene cause myostatin-related muscle hypertrophy. The *MSTN* gene provides instructions for making a protein called myostatin, which is active in muscles used for movement (skeletal muscles) both before and after birth. This protein normally limits muscle growth, ensuring that muscles do not grow too large. Variants that reduce the production of functional myostatin lead to an overgrowth of muscle tissue.

[Learn more about the gene associated with Myostatin-related muscle hypertrophy](#)

- *MSTN*

Inheritance

Myostatin-related muscle hypertrophy has a pattern of inheritance known as incomplete autosomal dominance. People with a variant in both copies of the *MSTN* gene in each cell (homozygotes) have significantly increased muscle mass and strength. People with a variant in one copy of the *MSTN* gene in each cell (heterozygotes) also have increased muscle bulk, but to a lesser degree.

Other Names for This Condition

- Muscle hypertrophy syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Myostatin-related muscle hypertrophy (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2931112/>)

Genetic and Rare Diseases Information Center

- Myostatin-related muscle hypertrophy (<https://rarediseases.info.nih.gov/diseases/10238/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

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

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28myostatin+AND+muscle+hypertrophy%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

References

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- Schuelke M, Wagner KR, Stolz LE, Hubner C, Riebel T, Komen W, Braun T, TobinJF, Lee SJ. Myostatin mutation associated with gross muscle hypertrophy in achild. N Engl J Med. 2004 Jun 24;350(26):2682-8. doi: 10.1056/NEJMoa040933. Noabstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15215484>)

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