



Myostatin-related muscle hypertrophy

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

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 restrains muscle growth, ensuring that muscles do not grow too large. Mutations that reduce the production of functional myostatin lead to an overgrowth of muscle tissue.

Inheritance Pattern

Myostatin-related muscle hypertrophy has a pattern of inheritance known as incomplete autosomal dominance. People with a mutation in both copies of the *MSTN* gene in each cell (homozygotes) have significantly increased muscle mass and strength. People with a mutation 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

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
</primer/testing/genetictesting>
- Genetic Testing Registry: Myostatin-related muscle hypertrophy
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2931112/>

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Muscle Disorders
<https://medlineplus.gov/muscledisorders.html>

Genetic and Rare Diseases Information Center

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

Educational Resources

- MalaCards: myostatin-related muscle hypertrophy
https://www.malacards.org/card/myostatin_related_muscle_hypertrophy
- Neuromuscular Disease Center, Washington University
<https://neuromuscular.wustl.edu/mother/mlarge.html#myostatinmut>
- Orphanet: Myostatin-related muscle hypertrophy
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=275534

Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/muscular.html>

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- MYOSTATIN
<http://omim.org/entry/601788>

Medical Genetics Database from MedGen

- Myostatin-related muscle hypertrophy
<https://www.ncbi.nlm.nih.gov/medgen/418994>

Sources for This Summary

- Carnac G, Ricaud S, Vernus B, Bonnieu A. Myostatin: biology and clinical relevance. *Mini Rev Med Chem.* 2006 Jul;6(7):765-70. Review.
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 - Schuelke M, Wagner KR, Stolz LE, Hübner C, Riebel T, Kömen W, Braun T, Tobin JF, Lee SJ. Myostatin mutation associated with gross muscle hypertrophy in a child. *N Engl J Med.* 2004 Jun 24;350(26):2682-8.
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