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
Other Diagnosis and Management Resources
- GeneReview: Myostatin-Related Muscle Hypertrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1498

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

Clinical Information from GeneReviews
- Myostatin-Related Muscle Hypertrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1498

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+AND+%225Bdp%5D

Catalog of Genes and Diseases from OMIM
- MYOSTATIN
  http://omim.org/entry/601788
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16842126

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15215484

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301671

Reprinted from Genetics Home Reference:

Reviewed: December 2008
Published: November 7, 2018

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services