Grange syndrome

Description

Grange syndrome is a rare condition that primarily affects the blood vessels. It is characterized by narrowing (stenosis) or blockage (occlusion) of arteries that supply blood to various organs and tissues, including the kidneys, brain, and heart. Stenosis or occlusion of the arteries that supply blood to the kidneys (renal arteries) can result in chronic high blood pressure (hypertension). Blockage of the arteries that carry blood to the brain (cerebral arteries) can cause a stroke.

Additional features of Grange syndrome can include short fingers and toes (brachydactyly), fusion of some of the fingers or toes (syndactyly), fragile bones that are prone to breakage, and learning disabilities. Most people with this disorder also have heart defects that are present from birth.

Frequency

Grange syndrome has been reported to affect at least six individuals from three families.

Causes

Grange syndrome results from mutations in the *YY1AP1* gene. The protein produced from this gene is part of a group of proteins (a complex) that helps regulate several critical functions within cells. These include gene activity (expression), repair of damaged DNA, cell specialization (differentiation), and cell growth and division (proliferation). Researchers believe that this protein complex plays a particularly important role in smooth muscle cells, which line the walls of blood vessels.

Mutations in the *YY1AP1* gene likely disrupt the function of the complex, which leads to reduced proliferation and differentiation of smooth muscle cells. However, it is unclear how these changes lead to narrowing and blockage of arteries. It is also unknown how *YY1AP1* gene mutations are related to other features of Grange syndrome, such as bone abnormalities and learning disabilities.

Learn more about the gene associated with Grange syndrome

- YY1AP1

Inheritance

This condition is thought to be inherited in an autosomal recessive pattern, which means both copies of the *YY1AP1* gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Arterial occlusive disease, progressive, with hypertension, heart defects, bone fragility, and brachysyndactyly
- Grange occlusive arterial syndrome
- GRNG

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Grange syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1865267/)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- GRANGE SYNDROME; GRNG (https://omim.org/entry/602531)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Grange+syndrome%5BTIAB%5D%29+OR+%28%28arterial+occlusive+disease%5BTIAB%5D%29+AND+%28brachydactyly%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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Last updated July 1, 2017