Complement factor I deficiency

Complement factor I deficiency is a disorder that affects the immune system. People with this condition are prone to recurrent infections, including infections of the upper respiratory tract, ears, skin, and urinary tract. They may also contract more serious infections such as pneumonia, meningitis, and sepsis, which may be life-threatening.

Some people with complement factor I deficiency have a kidney disorder called glomerulonephritis with isolated C3 deposits. Complement factor I deficiency can also be associated with autoimmune disorders such as rheumatoid arthritis or systemic lupus erythematosus (SLE). Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs.

Frequency

Complement factor I deficiency is a rare disorder; its exact prevalence is unknown. At least 38 cases have been reported in the medical literature.

Causes

Complement factor I deficiency is caused by mutations in the CFI gene. This gene provides instructions for making a protein called complement factor I. This protein helps regulate a part of the body's immune response known as the complement system. The complement system is a group of proteins that work together to destroy foreign invaders (such as bacteria and viruses), trigger inflammation, and remove debris from cells and tissues. This system must be carefully regulated so it targets only unwanted materials and does not attack the body's healthy cells. Complement factor I and several related proteins protect healthy cells by preventing activation of the complement system when it is not needed.

Mutations in the CFI gene that cause complement factor I deficiency result in abnormal, nonfunctional, or absent complement factor I. The lack (deficiency) of functional complement factor I protein allows uncontrolled activation of the complement system. The unregulated activity of the complement system decreases blood levels of another complement protein called C3, reducing the immune system's ability to fight infections. In addition, the immune system may malfunction and attack its own tissues, resulting in autoimmune disorders.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the 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

- C3 inactivator deficiency
- complement component 3 inactivator deficiency
- hereditary factor I deficiency disease

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Hereditary factor I deficiency disease

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22complement+factor+I+deficiency%22+OR+%22complement+deficiency%22

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Complement
  https://medlineplus.gov/ency/article/003456.htm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Complement
  https://medlineplus.gov/ency/article/003456.htm
- Encyclopedia: Immunodeficiency Disorders
  https://medlineplus.gov/ency/article/000818.htm
- Health Topic: Immune System and Disorders
  https://medlineplus.gov/immunesystemanddisorders.html

Additional NIH Resources

- National Institute of Allergy and Infectious Diseases: Primary Immune Deficiency Diseases
Educational Resources

• MalaCards: complement factor i deficiency
  https://www.malacards.org/card/complement_factor_i_deficiency

• The Merck Manual for Healthcare Professionals: Complement System

Patient Support and Advocacy Resources

• Jeffrey Modell Foundation
  http://www.info4pi.org/

Scientific Articles on PubMed

• PubMed

Catalog of Genes and Diseases from OMIM

• COMPLEMENT FACTOR I DEFICIENCY
  http://omim.org/entry/610984

Sources for This Summary

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