

Complement Factor I Deficiency

Subjects: **Genetics & Heredity**

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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.

genetic conditions

1. Introduction

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.

2. 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.

3. 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.

4. The Gene Associated with Complement Factor I Deficiency

- CFI

5. Inheritance

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.

6. Other Names for This Condition

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

References

1. Baracho GV, Nudelman V, Isaac L. Molecular characterization of homozygous hereditary factor I deficiency. *Clin Exp Immunol.* 2003 Feb;131(2):280-6.
2. Grumach AS, Leitão MF, Arruk VG, Kirschfink M, Condino-Neto A. Recurrent infections in partial complement factor I deficiency: evaluation of three generations of a Brazilian family. *Clin Exp Immunol.* 2006 Feb;143(2):297-304.
3. Nilsson SC, Trouw LA, Renault N, Miteva MA, Genel F, Zelazko M, Marquart H, Muller K, Sjöholm AG, Truedsson L, Villoutreix BO, Blom AM. Genetic, molecular and functional analyses of complement factor I deficiency. *Eur J Immunol.* 2009 Jan;39(1):310-23. doi: 10.1002/eji.200838702.
4. Pettigrew HD, Teuber SS, Gershwin ME. Clinical significance of complement deficiencies. *Ann N Y Acad Sci.* 2009 Sep;1173:108-23. doi:10.1111/j.1749-6632.2009.04633.x. Review.
5. Ponce-Castro IM, González-Rubio C, Delgado-Cerviño EM, Abarregui-Garrido C, Fontán G, Sánchez-Corral P, López-Trascasa M. Molecular characterization of Complement Factor I deficiency in two Spanish families. *Mol Immunol.* 2008 May;45(10):2764-71. doi: 10.1016/j.molimm.2008.02.008.
6. Vyse TJ, Morley BJ, Bartok I, Theodoridis EL, Davies KA, Webster AD, Walport MJ. The molecular basis of hereditary complement factor I deficiency. *J Clin Invest.* 1996 Feb 15;97(4):925-33.
7. Vyse TJ, Späth PJ, Davies KA, Morley BJ, Philippe P, Athanassiou P, Giles CM, Walport MJ. Hereditary complement factor I deficiency. *QJM.* 1994 Jul;87(7):385-401. Review.

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