MBL2 Gene

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Mannose binding lectin 2

genes

1. Introduction

The *MBL2* gene provides instructions for making a protein that assembles into a protein complex called mannose-binding lectin. Functional mannose-binding lectins are made up of two to six protein groups called trimers, which are each composed of three of the protein pieces (subunits) produced from the *MBL2* gene. This protein complex plays an important role in the immune system's response to foreign invaders (pathogens).

Mannose-binding lectin recognizes and attaches (binds) to sugars, such as mannose, fucose, and glucose, that are found on the surface of bacteria, viruses, and yeast. This binding turns on (activates) the complement system, which is a group of immune system proteins that work together to destroy pathogens, trigger inflammation, and remove debris from cells and tissues. Attachment of mannose-binding lectin also targets the pathogen to be engulfed and broken down by special immune cells. Recognition of foreign invaders by mannose-binding lectin provides one of the body's first lines of defense against infection.

2. Health Conditions Related to Genetic Changes

2.1. Mannose-Binding Lectin Deficiency

Several common mutations of the *MBL2* gene can lead to a condition called mannose-binding lectin deficiency. People with this condition have low levels of mannose-binding lectin and may be susceptible to recurrent infections. Several of the disease-associated mutations occur in a region of the *MBL2* gene known as exon 1 and result in a change to single protein building blocks (amino acids) in the mannose-binding lectin subunit. Other mutations occur in an area of DNA near the *MBL2* gene called the promoter region, which helps control the production of the mannose-binding lectin subunit.

The change of a single amino acid in the mannose-binding lectin subunit eliminates its ability to assemble into the functional mannose-binding lectin. Similarly, certain mutations in the promoter region of the *MBL2* gene reduce production of the mannose-binding lectin subunit, leading to a decreased number of subunits available for protein assembly and a reduction in the amount of functional protein. With decreased levels of mannose-binding lectin, the

body does not recognize and fight foreign invaders efficiently. Consequently, infections can be more common in people with this condition. However, researchers believe that a number of factors, including other genetic and environmental factors, are involved in the development of mannose-binding lectin deficiency.

3. Other Names for This Gene

- COLEC1
- collectin-1
- HSMBPC
- mannan-binding lectin
- mannose-binding lectin (protein C) 2, soluble
- mannose-binding lectin (protein C) 2, soluble (opsonic defect)
- mannose-binding lectin 2, soluble (opsonic defect)
- mannose-binding protein C
- mannose-binding protein C precursor
- MBL
- MBL2_HUMAN
- MBL2D
- MBP
- MBP-C
- MBP1

References

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