CIITA Gene

Subjects: Genetics & Heredity
Contributor: Vicky Zhou

class II major histocompatibility complex transactivator

Keywords: genes

1. Normal Function

The *CIITA* gene provides instructions for making a protein that primarily helps control the activity (transcription) of genes called major histocompatibility complex (MHC) class II genes. Transcription is the first step in the production of proteins, and CIITA is critical for the production of specialized immune proteins called MHC class II proteins from these genes. The CIITA protein coordinates various proteins to turn on MHC class II gene transcription and allow the production of MHC class II proteins.

MHC class II proteins are found on the surface of several types of immune cells, including white blood cells (lymphocytes) that are involved in immune reactions. These proteins play an important role in the body's immune response to foreign invaders, such as bacteria, viruses, and fungi. To help the body recognize and fight infections, MHC class II proteins bind to fragments of proteins (peptides) from foreign invaders so that other specialized immune system cells can interact with them. When these immune system cells recognize the peptides as harmful, they trigger the lymphocytes and other immune cells to launch immune responses to get rid of the foreign invaders.

The CIITA protein also appears to play a role in enhancing the transcription of MHC class I genes, which provide instructions for making immune system proteins called MHC class I proteins. Like MHC class II proteins, MHC class I proteins attach to peptides from foreign invaders and present them to specific immune system cells. These cells then attack the foreign invaders to rid them from the body. While the CIITA protein is able to help promote MHC class I gene activity, it is not the primary regulator of these genes. Other proteins play a more prominent role in their transcription.

2. Health Conditions Related to Genetic Changes

2.1. Bare Lymphocyte Syndrome Type II

More than a dozen mutations in the *CIITA* gene have been found to cause an immune system disorder called bare lymphocyte syndrome type II (BLS II). BLS II is a type of combined immunodeficiency (CID), in which affected individuals have virtually no immune protection from foreign invaders. Consequently, individuals with BLS II have persistent infections in the respiratory, gastrointestinal, and urinary tracts, which can be life-threatening.

CIITA gene mutations involved in BLS II result in a lack of functioning CIITA protein. A shortage of CIITA to coordinate the factors that turn on MHC class II gene transcription prevents production of MHC class II proteins. Consequently, lymphocytes and other immune cells lack any MHC class II proteins on their surface, and the body has difficulty getting rid of bacteria, viruses, and fungi, leading to the persistent infections characteristic of BLS II.

2.2. Autoimmune Addison Disease

Autoimmune Addison disease

3. Other Names for This Gene

- C2TA
- CIITAIV
- MHC class II transactivator
- MHC2TA

- NLR family, acid domain containing
- NLRA
- nucleotide-binding oligomerization domain, leucine rich repeat and acid domain containing

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