Bare Lymphocyte Syndrome Type I

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Bare lymphocyte syndrome type I (BLS I) is an inherited disorder of the immune system (primary immunodeficiency). Immunodeficiencies are conditions in which the immune system is not able to protect the body effectively from foreign invaders such as bacteria or viruses. Starting in childhood, most people with BLS I develop recurrent bacterial infections in the lungs and airways (respiratory tract). These recurrent infections can lead to a condition called bronchiectasis, which damages the passages leading from the windpipe to the lungs (bronchi) and can cause breathing problems.

genetic conditions

1. Introduction

Many people with BLS I also have open sores (ulcers) on their skin, usually on the face, arms, and legs. These ulcers typically develop in adolescence or young adulthood. Some people with BLS I have no symptoms of the condition.

People with BLS I have a shortage of specialized immune proteins called major histocompatibility complex (MHC) class I proteins on cells, including infection-fighting white blood cells (lymphocytes), which is where the condition got its name.

2. Frequency

BLS I is a rare disorder with an unknown prevalence. About 30 affected individuals have been described in the medical literature. The condition is likely underdiagnosed, because doctors may not investigate the underlying cause of respiratory tract infections.

3. Causes

BLS I is usually caused by mutations in the *TAP1* or *TAP2* gene. Each of these genes provides instructions for making a protein that plays a role in helping the immune system recognize and fight infections. In particular, the TAP1 and TAP2 proteins aid the function of MHC class I proteins.

The TAP1 and TAP2 proteins attach (bind) together to form a protein complex called transporter associated with antigen processing (TAP) complex. This complex, which is found in the membrane of a cell structure called the

endoplasmic reticulum, moves (transports) protein fragments (peptides) from foreign invaders into the endoplasmic reticulum. There, the peptides are attached to MHC class I proteins. The peptide-bound MHC class I proteins are then moved to the surface of the cell so that specialized immune system cells can interact with them. When these immune system cells recognize the peptides as harmful, they launch an immune response to get rid of the foreign invaders.

Mutations in the *TAP1* or *TAP2* gene prevent formation of the TAP complex, which impairs the transport of peptides into the endoplasmic reticulum. Because there are no peptides for MHC class I proteins to bind, they are broken down, which results in a shortage of MHC class I proteins on the surface of cells. A lack of these proteins impairs the body's immune response to bacteria, leading to recurrent bacterial infections. Researchers are unsure why people with BLS I do not also get viral infections, but they suspect that other immune processes are able to recognize and fight viruses. It is also not clear how *TAP1* and *TAP2* gene mutations are involved in the development of skin ulcers.

Mutations in another gene involved in the attachment of peptides to MHC class I proteins very rarely cause BLS I.

3.1. The genes associated with Bare lymphocyte syndrome type I

- TAP1
- TAP2

3.2. Additional Information from NCBI Gene

- TAPBP

4. 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.

5. Other Names for This Condition

- HLA class I deficiency

References

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