Hyper IgM Syndrome

Hyper IgM syndrome is a congenital B-cell immunodeficiency caused by a spectrum of defects that ultimately result in an inability for B-cells to undergo class switching. Class switching describes the transformation of IgM antibodies to other classes e.g. IgG, IgE, IgA. Without this transformation, IgM antibodies accumulate. Examples of defects include deficient CD40 ligand on T-cells (type 1) and defective/deficient CD40 receptor on B-cells (type 3). Sometimes, the mutation is unknown. Without the diverse repertoire of immunoglobulins, patients with Hyper IgM syndrome are more prone to developing pyogenic abscesses and other infections. Serum levels of IgA, IgE, and IgG are decreased while IgM levels are elevated.



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Pathophysiology

Deficient CD40 Ligand on Helper T-cells

Broken CD-40 oz with Light-bulb on Tennis-ball

CD40 ligand on helper T-cells attaches to the CD40 receptor on B-cells, working to activate B-cells to undergo class switching and produce specific immunoglobulins for pathogen defense. In this syndrome, patients may have a defective CD40 ligand gene resulting in insufficient CD40 ligand production. Consequently, B-cells are unable to mount a specific immunoglobulin response and continue to produce non-specific IgM.

Defective CD40 Receptor on B-cells

CD-40 oz receptor on Basketball

Another pathogenetic process that results in Hyper IgM syndrome involves a defective CD40 receptor gene resulting in insufficient/poorly formed CD40 receptors. Consequently, B-cells are unable to mount a specific immunoglobulin response and continue to produce non-specific IgM.

Inability to Class Switch

Broken Glass Switch

Class switching describes the cell's ability to change immunoglobulin response from non-specific IgM to specific types of IgA, IgE, and IgG class immunoglobulins. In hyper IgM syndrome, there is defective class switching.

Signs and Symptoms

Pyogenic Abscess

Pus-pies and Abscess-guy

Patients with hyper IgM syndrome present with multiple abscesses, pyogenic infections, and neutropenia due to the inability to class switch. These typically present early in life.

Decreased IgG, IgA, IgE

Down-arrow Gold, Apple, Electric Globulin-Goblins

Patients without the ability to class switch are limited in their ability to produce specific immunoglobulin classes; such as IgA, IgE, and IgG. This results in an overproduction of non-specific IgM.

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