Wiskott Aldrich

Wiskott-Aldrich syndrome is a rare X-linked recessive disorder characterized by the triad of thrombocytopenic purpura, recurrent infections, and eczema. This disease is caused by a mutation in the WAS gene, which encodes the Wiskott-Aldrich syndrome protein (WASp). This protein plays a role in actin polymerization. In T cells, this protein is important because it is known to be activated by a T cell receptor signaling pathway to induce actin rearrangements that are responsible for the interface between an antigen presenting cell and a lymphocyte. A defective protein causes defective antigen presentation, leading to progressive deletion of both B and T cells. The first signs of the disease are often purpura and bruising caused by thrombocytopenia. Infants also develop eczema within the first month of life and develop recurrent infections by three months. Lab findings typically demonstrate decreased or normal IgG and IgM levels and increased IgA and IgE levels.

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Pathophysiology

X-linked Recessive

X-suit with Recessive-chocolate

This disease is inherited in an X linked recessive fashion. Due to its mode of inheritance, the majority of patients are male.

WASp Protein Mutation

Mutated Wasp

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Antigen Presentation Defect

Ant-gem Presentation Defected

Rearrangements of actin result in a defect of antigen presentation.

Progressive Deletion of B and T Cells

Deletion of Basketballs and Tennis-balls

Defect in actin polymerization leads to defective interface between antigen presenting cells and lymphocytes, leading to a progressive deletion of B and T cells.

Signs and Symptoms

Thrombocytopenia

Trombone-side-toe-peanut

The first signs of the disease are often bruising and bleeding caused by thrombocytopenia.

Eczema

Ax-zebra

Infants commonly develop eczema within the first month of life, possibly related to the elevated IgE levels leading to an increase in atopy.

Infections

Bacteria, Virus, Fungus, and Protozoa

Due to the progressive deletion of B and T cells, recurrent infections are typically seen by three months of age.

Decreased IgM

Murdered Down-arrow (IgM) Mountain-goblin

Lab findings typically demonstrate low IgM levels, with elevated IgA and IgE.

Increased IgE, IgA

Up-arrow Electric and Apple Globulin-Goblins

Lab findings typically demonstrate low IgM levels, with elevated IgA and IgE.