







# IMMUNE DEFICIENCY DISEASES







Immunodeficiency disorders are a group of illnesses caused by deficient cells.

#### Classification

Primary immunodeficiency: Inherited disorders due to genetic mutations, manifesting at birth or during childhood.

#### **DiGeorge Syndrome**

Defect in the development of Thymus and Parathyroid glands resulting in T-cell deficiency.

#### **Pathogenesis**

- Deletion of chromosome 22q11 leads to failure of development of the 3rd and 4th pharyngeal pouch which are responsible for the development of Thymus & Parathyroid gland.
- T-cell defect leads to increased infection by intracellular pathogens.





### **Defects**



- Abnormal facies
- Thymic aplasia
- Cleft palate
- Hypocalcemia
- 22q11 chromosome deletion
- Tip to remember: These defects can be easily remembered by a famous mnemonic: CATCH 22.





# Bruton's agammaglobulinemia

- Also known as X-linked agammaglobulinemia.
- X-linked disorder is characterized by a defect in B cell development defect.

#### **Pathogenesis:**

- Defect in B cell tyrosine kinase leads to the production of abnormal immunoglobulins resulting in their deficiency, hence the name agammaglobulinemia.
- B cell defect leads to increased infection by extracellular pathogens.

#### **Presentation:**

- Presents after 6 months of age
- Recurrent infections.

#### **Tip to remember: Bruton uses X (Twitter)**

- B for Bruton's disease, caused by B cells.
- X for X-linked & extracellular.







# Common variable immunodeficiency (CVID)

Characterized by defective antibody production.

#### **Pathogenesis:**

• A defect in B cell maturation leads to defective antibody production.

#### **Presentation:**

- Can present in any age.
- Recurrent infections.
- Increased risk of Autoimmune diseases
- Malignancies

Tip to remember: it can be remembered as the less severe version of Bruton, so Bruton ka Chhota bhai CVID.







# Severe Combined Immunodeficiency (SCID)

Characterized by severe impairment of both B cells & T cells.

#### **Pathogenesis:**

- Enzyme deficiency: adenosine deaminase deficiency, shows autosomal recessive inheritance.
- Defect in cytokine receptors shows X-lined inheritance

#### **Presentation:**

- Early onset
- Strong susceptibility to life-threatening infections.

Tips to remember: the name itself suggests a combined deficiency of both B & T cells. A for ADA enzyme & autosomal dominant.







# Secondary immunodeficiency

Acquired due to infection, drugs, etc.

**Human Immunodeficiency Virus (HIV) infection** 

Causes immunodeficiency leading to increased susceptibility to infections and malignancies.

#### **Pathogenesis:**

- HIV infects CD 4 T helper cells and macrophages.
- Results in depletion of CD4 T helper cells causing progressive loss of immunity.





### **Etiology**



- Multiple sex partners
- Sex workers
- Homosexual intercourse
- 2. Blood and blood products
- Blood transfusion
- IV drug users
- Accidental exposure to Health workers
- 3. Vertical transmission:
- from mother to child
- during childbirth or breastfeeding
- However, HIV infection in mother is not an absolute contraindication of breastfeeding in India.



### **Clinical Presentation**

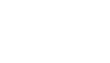




- Acute HIV infection
- Chronic asymptomatic HIV infection
- Chronic symptomatic HIV infection
- AIDS: Acquired Immune Deficiency Syndrome
- CD4 counts <200 cells/ul
- Opportunistic infections like cytomegalovirus, tuberculosis.
- AIDS Defining Malignancies like Kaposi sarcoma, Non-Hodgkin Lymphoma, and Cervical cancer.



### Tips to remember



- H for HIV, Helper T cells.
- All high-risk groups are commonly known, like IV drug users sharing needles, sex workers, truck drivers, etc.
- 1. Immunosuppressive drugs
- Corticosteroids
- Chemotherapeutic agents
- 2. Malnutrition
- Each of these disorders can lead to serious implications like recurrent
- infections, failure to thrive, or increased risk of autoimmune disorders and malignancies.

