

#MADEEASY



**IMMUNE DEFICIENCY
DISEASES**





Immunodeficiency

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



- **Congenital cardiac abnormalities**
- **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



1. **Sexual contact**
 - **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/ μ l**
- **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.*

