



## Genetic and Acquired Immunodeficiencies: Causes and Treatments

Mutsa Takundwa\*

Department of Medicine, University of Oklahoma College of Medicine, Oklahoma, United States of America

### DESCRIPTION

Immunodeficiencies represent a group of disorders where the immune system's ability to fight infectious diseases and cancer is compromised or entirely absent. These conditions can be broadly categorized into genetic (primary) and acquired (secondary) immunodeficiencies. Both types result in increased susceptibility to infections, autoimmune diseases, and malignancies, but they differ in their etiology and, often, in their treatment approaches. Genetic immunodeficiencies, also known as Primary Immunodeficiency Diseases (PIDs), are usually inherited and present at birth. They result from mutations in genes that are essential for the development and function of the immune system. There are over 400 different types of PIDs, each with varying degrees of severity.

One of the most well-known genetic immunodeficiencies is Severe Combined Immunodeficiency (SCID), often termed "bubble boy disease." SCID is characterized by a severe defect in both T and B lymphocyte function, leading to a profound vulnerability to infections. Patients with SCID require a sterile environment to prevent infections and often succumb to these infections if untreated. Other examples of PIDs include Common Variable Immunodeficiency (CVID), which typically presents later in childhood or adulthood with recurrent infections and autoimmune disorders, and Chronic Granulomatous Disease (CGD), which is caused by defects in the ability of phagocytes to kill ingested pathogens.

Genetic immunodeficiencies are caused by mutations in specific genes responsible for the immune system's development and function. These mutations can be inherited in various patterns, including autosomal dominant, autosomal recessive, and X-linked recessive. SCID can be caused by mutations in several genes. The underlying genetic defects disrupt normal immune cell development, signaling, or function, leading to immunodeficiency. Treatment strategies for genetic immunodeficiencies focus on restoring immune function and

preventing infections. Hematopoietic Stem Cell Transplantation (HSCT) is a curative treatment for many severe PIDs, such as SCID. HSCT involves replacing the patient's defective immune system with healthy immune cells from a compatible donor. For milder forms of PIDs, treatments may include immunoglobulin replacement therapy, which provides the patient with antibodies to help prevent infections, and prophylactic antibiotics to reduce the risk of bacterial infections.

One of the most significant acquired immunodeficiencies is Acquired Immunodeficiency Syndrome (AIDS), caused by the Human Immunodeficiency Virus (HIV). HIV targets and destroys T-cells, an essential component of the immune system, leading to severe immunodeficiency over time. Other causes of acquired immunodeficiencies include chemotherapy and radiation therapy for cancer, which can damage the bone marrow and impair immune function, and immunosuppressive drugs used to prevent organ transplant rejection or treat autoimmune diseases. The causes of acquired immunodeficiencies are diverse. HIV/AIDS is the most prominent example, but other infections, like certain viral, bacterial, and parasitic infections, can also suppress the immune system. Malnutrition, particularly protein-calorie malnutrition, and deficiencies in specific vitamins and minerals can impair immune function. Chemotherapy, radiation therapy, and immunosuppressive drugs can significantly weaken the immune system. Conditions like diabetes, renal failure, and chronic liver disease can lead to secondary immunodeficiency.

Treatment of acquired immunodeficiencies involves addressing the underlying cause and supporting the immune system. For HIV/AIDS, Antiretroviral Therapy (ART) is used to suppress the virus and restore immune function. Patients undergoing chemotherapy or receiving immunosuppressive drugs may be given growth factors to stimulate white blood cell production, prophylactic antibiotics, or antifungal agents to prevent infections. Nutritional support and supplements can help improve immune function in malnourished individuals.

**Correspondence to:** Mutsa Takundwa, Department of Medicine, University of Oklahoma College of Medicine, Oklahoma, United States of America, E-mail: mutsa@pathol.edu

**Received:** 21-May-2024, Manuscript No. BLM-24-26251; **Editor assigned:** 23-May-2024, PreQC No. BLM-24-26251 (PQ); **Reviewed:** 07-Jun-2024, QC No. BLM-24-26251; **Revised:** 14-Jun-2024, Manuscript No. BLM-24-26251 (R); **Published:** 21-Jun-2024, DOI: 10.35248/0974-8369.24.16.698.

**Citation:** Takundwa M (2024) Genetic and Acquired Immunodeficiencies: Causes and Treatments. *Bio Med.* 16:698.

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