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**Nursing Science**

**NSC 306**

**Primary Immunodeficiency Disorders**:

APS-1 (APECED):-

Autoimmune polyglandular syndrome type 1 (APS-1), also called autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED), is a genetic immune disorder. This disorder has a diverse range of symptoms, including autoimmunity against different organs and an increased susceptibility to candidiasis, a fungal infection caused by Candida yeast.

The syndrome is caused by mutations in the autoimmune regulator (AIRE) gene. NIAID researchers are exploring how AIRE mutations impact the function of cells by studying people with APS-1 (APECED), as well as using mouse models of AIRE deficiency. With a better understanding of how these mutations affect the immune system and other cells in the body, researchers hope to uncover promising therapeutic targets to prevent and treat persistent candidiasis and autoimmune disorders.

CEDS:-

CEDS is a very rare genetic disorder of the immune system. CEDS stands for “caspase eight deficiency state.” The disorder is characterized by an enlarged spleen and lymph nodes, recurrent sinus and lung infections, recurrent viral infections, and a low level of infection-fighting antibodies. CEDS is diagnosed based on clinical and laboratory findings as well as genetic testing.

CEDS is caused by mutations in the CASP8 gene, which provides instructions for production of the protein caspase eight, which is also abbreviated as CASP8. The CASP8 protein is involved in programmed cell death, or apoptosis. The body must maintain a careful balance between proliferation of immune cells and apoptosis to defend against pathogens and avoid autoimmunity. The mutations that cause CEDS destabilize the CASP8 protein and block its function, leading to buildup of immune cells.

Chronic Granulomatous Disease (CGD):-

Chronic granulomatous disease (CGD) is a genetic disorder in which white blood cells called phagocytes are unable to kill certain types of bacteria and fungi. People with CGD are highly susceptible to frequent and sometimes life-threatening bacterial and fungal infections. CGD is caused by defects in an enzyme, NADPH oxidase, that phagocytes need to kill certain bacteria and fungi. Mutations in one of five different genes can cause these defects. People with CGD are highly susceptible to infections caused by certain bacteria and fungi, such as Staphylococcus aureus, Serratia marcescens, Burkholderia cepacia, Nocardia species, and Aspergillus species. These people may develop abscesses (boils) in their lungs, liver, spleen, bones, or skin; and masses of cells, called granulomas, that can obstruct the bowel or urinary tract. In some people, granulomas can cause an inflammatory bowel disease similar to Crohn’s Disease. In addition, heart or kidney problems, diabetes, and autoimmune disease may occur in people with CGD, but this varies depending on which gene is mutated.

PI3 Kinase Disease:-

PI3 kinase (PI3K) disease is a rare disorder that severely impairs the immune system’s ability to fight bacterial and viral infections. The disease also increases a person’s risk of lymphoma, a type of immune cell cancer. PI3K disease is sometimes called PASLI disease (short for PI3K-p110δ activating mutation causing senescent T cells, lymphadenopathy, and immunodeficiency) or APDS (for activating PI3K delta syndrome).

PI3K disease is caused by mutations in the genes PIK3CD or PIK3R1, which provide instructions for producing a protein called PI3K-p110δ. These mutations can affect the immune system by overactivating an important immune system signaling pathway. This overactivation launches a chain reaction leading to disruptions in the normal development of B and T cells, which play a key role in fighting pathogens, and to increased susceptibility to infection.

PI3K disease is characterized by recurrent respiratory infections that can lead to progressive airway damage. People with PI3K disease have too few of some types of immune cells and too many of others. This sometimes includes a buildup of immune cells called lymphocytes, which can lead to swelling of the lymph nodes and spleen, chronic Epstein-Barr virus and cytomegalovirus infections, and an increased risk for lymphoma. Many people with PI3K disease also have abnormal levels of certain types of antibodies.

Warts, Hypogammaglobulinemia, Infections, and Myelokathexis (WHIM) Syndrome:-

Warts, Hypogammaglobulinemia, Infections, and Myelokathexis (WHIM) syndrome is a rare genetic disease of the immune system. Its name is an acronym for its main clinical manifestations: warts, hypogammaglobulinemia, infections, and myelokathexis. Hypogammaglobulinemia is a deficiency in specific infection-fighting antibodies in the blood. Myelokathexis refers to the failure of neutrophils infection-fighting white blood cells to move from the bone marrow into the bloodstream where they can patrol the body. WHIM syndrome patients also have trouble distributing most other types of immune cells to the blood. Such defects in the immune system predispose WHIM syndrome patients to frequent bacterial and viral infections, persistent skin and genital warts, and an increased risk of developing cancer caused by human papillomavirus.

**Secondary Immunodeficiency Disorders**:

HIV:-

HIV is a virus that damages the immune system. The immune system helps the body fight off infections. Untreated HIV infects and kills CD4 cells, which are a type of immune cell called T cells. Over time, as HIV kills more CD4 cells, the body is more likely to get various types of infections and cancers.

HIV is transmitted through bodily fluids that include: blood, semen, vaginal and rectal fluids and breast milk. The virus doesn’t spread in air or water, or through casual contact.

HIV is a lifelong condition and currently there is no cure, although many scientists are working to find one. However, with medical care, including treatment called antiretroviral therapy, it’s possible to manage HIV and live with the virus for many years.

Without treatment, a person with HIV is likely to develop a serious condition called AIDS. At that point, the immune system is too weak to fight off other diseases and infections. Untreated, life expectancy with AIDS is about three years. With antiretroviral therapy, HIV can be well-controlled and life expectancy can be nearly the same as someone who has not contracted HIV.

It’s estimated that 1.1 million Americans are currently living with HIV. Of those people, 1 in 5 don’t know they have the virus. HIV can cause changes throughout the body.

Multiple myeloma:-

Multiple myeloma is a type of cancer that affects plasma cells. Plasma cells are a type of white blood cell found in bone marrow, which is the soft tissue inside most of your bones that produces blood cells. In the bone marrow, plasma cells make antibodies. These are proteins that help your body fight off diseases and infections.

Multiple myeloma occurs when an abnormal plasma cell develops in the bone marrow and reproduces itself very quickly. The rapid reproduction of malignant, or cancerous, myeloma cells eventually outweighs the production of healthy cells in the bone marrow. As a result, the cancerous cells begin to accumulate in the bone marrow, crowding out the healthy white blood cells and red blood cells.

Like healthy blood cells, cancerous cells try to make antibodies. However, they can only produce abnormal antibodies called monoclonal proteins, or M proteins. When these harmful antibodies collect in the body, they can cause kidney damage and other serious problems.

Types of multiple myeloma:-

There are two main types of multiple myeloma. They’re categorized by their effect on the body:

1. An indolent myeloma causes no noticeable symptoms. It usually develops slowly and doesn’t cause bone tumors. Only small increases in M protein and M plasma cells are seen.
2. A solitary plasmacytoma causes a tumor to form, typically in bone. It usually responds well to treatment, but needs close monitoring.