Med-Surg Assignment

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1. Identify and briefly explain 5 primary immunodeficiency disorders

2. Identify and briefly explain 2 secondary immunodeficiency disorders.

1. i)Common Variable Immunodeficiency (CVID)

ii)Autoimmune Polyglandular Syndrome type 1 l,APS-1 (APECED).

iii) Benta Disease .

iv) Capase Eight Deficiency State.

v)Chronic Granulomatous

Common Variable Immunodeficiency (CVID):

Common variable immunodeficiency (CVID) is a primary immune deficiency disease characterized by low levels of protective antibodies and an increased risk of infections. Although the disease usually is diagnosed in adults, it also can occur in children. CVID also is known as hypogammaglobulinemia,adult-onset agammaglobulinemia,late-onsethypogammaglobulinemia, and acquired agammaglobulinemia. CVID is caused by a variety of different genetic abnormalities that result in a defect in the capability of immune cells to produce normal amounts of all types of antibodies. Only a few of these defects have been identified, and the cause of most cases of CVID is unknown. CVID is also linked to IgA deficiency, a related condition in which only the level of the antibody immunoglobulin A (IgA) is low, while levels of other antibody types are usually normal or near normal. IgA deficiency typically occurs alone, but in some cases it may precede the development of CVID or occur in family members of CVID patients.

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.

BENTA Disease:

BENTA disease is a rare genetic disorder of the immune system caused by mutations in the gene CARD11. The disease is characterized by high levels of certain immune cells starting in infancy, an enlarged spleen, enlarged lymph nodes, immunodeficiency, and an elevated risk of lymphoma, a type of cancer. BENTA disease is a rare genetic disorder of the immune system caused by mutations in the gene CARD11. BENTA stands for “B-cell expansion with NF-jB and T-cell anergy.” The disease is characterized by high levels of certain immune cells starting in infancy, an enlarged spleen, enlarged lymph nodes, immunodeficiency, and an increased risk of lymphoma, a type of cancer. Breaking down the name, BENTA, helps explain the syndrome.

• A B cell is a type of immune cell from the bone marrow.

• Expansion means that the number of B cells is greater than normal.

• NF-jB is a protein complex involved in gene expression, or the degree to which certain genes are turned on or off.

• A T cell is a type of immune cell that matures in the thymus, a small organ located in the upper chest under the breastbone.

• Anergy refers to a “less-than-normal” (T cell) immune reaction to foreign substances. BENTA disease is diagnosed based on clinical and laboratory findings as well as genetic testing. BENTA disease is caused by “gain- of-function” mutations in the gene CARD11, which provides instructions for production of the CARD11 pro- tein. These gain-of-function mutations cause the CARD11 protein to be overactive. The CARD11 protein is required for activation of NF-jB in both B and T cells, which is essen- tial for a healthy immune response. The development and differentiation of B cells might also be partially impaired in BENTA disease.

Caspase Eight Deficiency State (CEDS):

Caspase eight deficiency state, or CEDS, is a very rare genetic disorder of the immune system caused by mutations in the CASP8 gene. CEDS 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 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.

2. i)HIV/AIDS

ii)Leukemia

HIV/AIDS: HIV (human immunodeficiency virus) is a virus that attacks cells that help the body fight infection, making a person more vulnerable to other infections and diseases. It is spread by contact with certain bodily fluids of a person with HIV, most commonly during unprotected sex (sex without a condom or HIV medicine to prevent or treat HIV), or through sharing injection drug equipment.

If left untreated, HIV can lead to the disease AIDS (acquired immunodeficiency syndrome). AIDS is the late stage of the infection that occurs when the body’s immune system is badly damaged because of the virus. If AIDS does develop, it means that the immune system is severely compromised. It’s weakened to the point where it can no longer fight off most diseases and infections.

Leukemia: A cancer of blood-forming tissues, hindering the body's ability to fight infection.Leukaemia is cancer of blood-forming tissues, including bone marrow. Many types exist such as acute lymphoblastic leukaemia, acute myeloid leukaemia and chronic lymphocytic leukaemia. Leukemia is a group of blood cancers that usually begin in the bone marrow and result in high numbers of abnormal blood cells.These blood cells are not fully developed and are called blast or leukemia cells . Acute leukemia develops quickly and worsens rapidly, but chronic leukemia gets worse over time. Leukemia develops when the DNA of developing blood cells, mainly white cells, incurs damage. This causes the blood cells to grow and divide uncontrollably.Healthy blood cells die, and new cells replace them. These develop in the bone marrow.The abnormal blood cells do not die at a natural point in their life cycle. Instead, they build up and occupy more space.

As the bone marrow produces more cancer cells, they begin to overcrowd the blood, preventing the healthy white blood cells from growing and functioning normally.

Eventually, the cancerous cells outnumber healthy cells in the blood.