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1. Identify and briefly explain 5 primary immunodeficiency disorders.
2. Identify and briefly explain 2 secondary immunodeficiency disorders.

Answer

1a. X-linked agammaglobulinemia: X-linked agammaglobulinemia (XLA) is a condition that affects the immune system and occurs almost exclusively in males. People with XLA have very few B cells, which are specialized white blood cells that help protect the body against infection. B cells can mature into the cells that produce special proteins called antibodies or immunoglobulins. Antibodies attach to specific foreign particles and germs, marking them for destruction. Individuals with XLA are more susceptible to infections because their body makes very few antibodies. It can also be called agammaglobulinemia, Bruton's agammaglobulinemia, congenital agammaglobulinemia, hypogammaglobulinemia.

Causes

Mutations in the BTK gene cause XLA. This gene provides instructions for making the BTK protein, which is important for the development of B cells and normal functioning of the immune system. Most mutations in the *BTK* gene prevent the production of any BTK protein. The absence of functional BTK protein blocks B cell development and leads to a lack of antibodies. Without antibodies, the immune system cannot properly respond to foreign invaders and prevent infection.

B. Common variable immunodeficiency: Common Variable Immunodeficiency (CVID) is an antibody deficiency that leaves the immune system unable to defend against bacteria and viruses, resulting in recurrent and often severe infections primarily affecting the ears, sinuses, and respiratory tract.  (sinopulmonary infections). In the majority of cases, the diagnosis is not made until the third to fourth decade of life. Permanent damage to the respiratory tract (bronchiectasis) may occur due to severe and repeated infections.

Although genetic mutations that lead to CVID have been identified, the exact cause and genetic inheritance pattern of CVID is unknown in most cases. Both males and females are affected. It is one of the most common forms of primary immunodeficiency disease (PIDD), and the severity of symptoms varies from one person with the disease to another. CVID can be associated with autoimmune disorders that affect other blood cells causing low numbers of white cells or platelets, anaemia, arthritis and other conditions such as endocrine disorders.  Gastrointestinal problems including chronic diarrhea, weight loss, nausea, vomiting and abdominal pain can also be present. In some forms of CVID, patients develop granulomas in the lungs, lymph nodes, liver, skin or other organs. People with CVID are also at an increased risk for certain cancers (lymphoid and gastrointestinal cancers primarily).

C. Severe combined immunodeficiency: Severe Combined Immunodeficiency (SCID) is an inherited primary immunodeficiency diseases (PIDD) that typically presents in infancy results in profound immune deficiency condition resulting in a weak immune system that is unable to fight off even mild infections. It is considered to be the most serious PIDD.

SCID is caused by genetic defects that affects the function of T cells. Depending on the type of SCID, B cells and NK cells can also be affected. These cells play important roles in helping the immune system battle bacteria, viruses and fungi that cause infections. Affected infants will often die within the first year of life without treatment with hematopoietic stem cell transplantation. (HSCT) New-born screening for SCID is able to identify infants before they get sick, leading to a shorter time to transplant and offering improved outcomes following transplantation. There are several forms of SCID. The most common type is linked to a problem in a gene on the X chromosome, affecting only males. Women may carry the condition, but they also inherit a normal X chromosome. Since starting new-born screening for SCID, recessive forms of the disease that can affect boys and girls have been identified with increased frequency.

D. Job syndrome: Hyperimmunoglobulinemia E syndrome (HIES), of which the autosomal dominant form is called Buckley syndrome, is a heterogeneous group of immune disorders. Job's is also very rare at about 300 cases currently in the literature.

Abnormal neutrophil chemotaxis due to decreased production of interferon gamma by T lymphocytes is thought to cause the disease.

E. BENTA: 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.

2a. HIV/AIDS: Human immunodeficiency virus infection and acquired immune deficiency syndrome (HIV/AIDS) is a spectrum of conditions caused by infection with the human immunodeficiency virus (HIV). Following initial infection a person may not notice any symptoms, or may experience a brief period of influenza-like illness. Typically, this is followed by a prolonged period with no symptoms. If the infection progresses, it interferes more with the immune system, increasing the risk of developing common infections such as tuberculosis, as well as other opportunistic infections, and tumors which are otherwise rare in people who have normal immune function. These late symptoms of infection are referred to as acquired immunodeficiency syndrome (AIDS). This stage is often also associated with unintended weight loss.

HIV is spread primarily by unprotected sex (including anal and oral sex), contaminated blood transfusion, hypodermic needles, and from mother to child,  during pregnancy, delivery, or breastfeeding. Some bodily fluids, such as saliva, sweat and tears, do not transmit the virus. HIV is a member of the group of viruses known as retroviruses.

Methods of prevention include safe sex, needle exchange program, treating those who are infected, pre and post exposure prophylaxis, and male circumcision. Disease in a baby can often be prevented by giving both the mother and child antiretroviral medications. There is no cure or vaccine, however, antiretroviral treatment can slow the course of the disease and may lead to a near-normal life expectancy. Treatment is recommended as soon as the diagnosis is made.

B. Multiple myeloma: Multiple myeloma is a cancer that forms in a type of white blood cell called a plasma cell. Plasma cells help you fight infections by making antibodies that recognize and attack germs. Multiple myeloma causes cancer cells to accumulate in the bone marrow, where they crowd out healthy blood cells. Rather than produce helpful antibodies, the cancer cells produce abnormal proteins that can cause complications.

Cause

It's not clear what causes myeloma.

Physicians know that myeloma begins with one abnormal plasma cell in your bone marrow, the soft, blood-producing tissue that fills in the centre of most of your bones. The abnormal cell multiplies rapidly. Because cancer cells don't mature and then die as normal cells do, they accumulate, eventually overwhelming the production of healthy cells. In the bone marrow, myeloma cells crowd out healthy white blood cells and red blood cells, leading to fatigue and an inability to fight infections. The myeloma cells continue trying to produce antibodies, as healthy plasma cells do, but the myeloma cells produce abnormal antibodies that the body can't use. Instead, the abnormal antibodies (monoclonal proteins, or M proteins) build up in the body and cause problems such as damage to the kidneys. Cancer cells can also cause damage to the bones that increases the risk of broken bones.