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**QUESTIONS**

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

**PRIMARY IMMUNODEFICIENCY DISODERS OR DISEASES**

It refers to a heterogeneous group of disorders characterized by poor or absent function in one or more components of the immune system. They are inherited defects of the immune system. These defects may be in specific or nonspecific immune mechanisms. They are classified on the basis of the site of lesion in the differentiation or developmental pathway of the immune system. They are also disorders in which part of the components of the immune system is missing or whose function is altered or diminished.

**The primary immunodeficiency disorders include;**

* **Immunoglobulin A deficiency**
* **Severe combined immunodeficiency disorders**
* **Thymic hypoplasia**
* **Granulomatoses with polyangiitis**
* **Combined variable immunodeficiency**

**IMMUNOGLOBULIN A DEFICIENCY:** It is a primary genetic immunodeficiency, a type of the hypogammaglobulinemia. People with this deficiency lack immunoglobulin A (IgA), a type of antibody that protects against infections of the mucous membranes lining in the mouth, airways and digestive tract. Selective immunoglobulin A deficiency is the most common of the primary antibody deficiencies. Total immunoglobulin A deficiency is defined as an undetectable serum immunoglobulin A level at a value less than 5mg/dL in humans. Partial immunoglobulin A deficiency refers to a detectable but decreased immunoglobulin A levels that are more than 2 standard deviation below the normal age adjusted means. Immunoglobulin A deficiency is associated with normal B lymphocytes in peripheral blood, normal CD4+ and CD8+ T cells, and usually, normal neutrophil and lymphocyte count.

Both IgA subclasses, IgA1 and IgA2, are usually markedly reduced or absent, although isolated deficiencies of each subclass have been described. In general, an IgA2 deficiency would be expected to lead to more serious infections than an IgA1 deficiency, due to the stronger protease susceptibility of the IgA1 molecule.

**GRANULOMATOSES WITH POLYANGIITIS**: previously known as Wegener’s Granulomatoses (WG) is an extremely rare long term systemic disorder that involves the formation of granulomas and inflammation of blood vessels (vasculitis). It is a form of vasculitis that affects small and medium sized vessels in many organs but most commonly affects the upper respiratory tract, lungs and kidneys. The signs and symptoms of GPA are highly varied and reflect which organs are supplied by the affected blood vessels. Typical signs and symptoms include nosebleeds, stuffy nose and crustiness of nasal secretions and inflammation of the uveal layer of the eye. Damage to the heart, kidneys and lungs could be fatal. The cause of GPA is unknown. Genetics have been found to play a role in GPA though the risk of inheritance appears to be low. GPA treatment depends on the severity of the disease. Severe disease is typically treated with a combination of immunosuppressive medications such as rituximab or cyclophosphamide and high dose corticosteroids to control the symptoms of the disease and Azathioprine, methotrexate to keep the disease under control Plasma exchange is also used in severe cases with damage to the lungs, kidneys or intestines.

**SEVERE COMBINED IMMUNODEFICIENCY DISORDERS:** it is rare genetic disorder characterized by the disturbed development of functional T cells and B cells caused by numerous genetic mutations that result in differing clinical presentations. SCID involves defective antibody response due to either direct involvement with B lymphocytes activation due to nonfunctional T-helper cells. Consequently, both arms (B cells and T cells) of the adaptive immune system are impaired due to a defect in one of several possible genes. SCID is the most severe form of primary Immunodeficiencies, and there are now at least nine different known genes in which mutations lead to form a SCID. It is also known as the bubble boy disease and bubble baby disease because its victims are extremely vulnerable to infectious diseases and some of them, such as David Vetter, have become famous for living in a sterile environment. SCID is the result of an immune system so highly compromised that it is considered almost absent. It is caused by genetic defects that affect 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. Common signs and symptoms of SCID include; and increased susceptibility to infections including ear infections, pneumonia or bronchitis, oral thrush and diarrhea. Due to recurrent infections, children with SCID do not grow and gain weight as expected (failure to thrive).

**THYMIC HYPOPLASIA:** this is a condition where the thymus is underdeveloped or involuted. Calcium levels can be used to distinguish between the following two conditions associated with thymic hypoplasia; 22q11.2 deletion syndrome; hypocalcaemia. Ataxia telangiectasia: normal levels of calcium.

**Ataxia-telangiectasia:** this is a rare neurodegenerative, autosomal recessive disease causing severe disability. Ataxia refers to poor coordination and telangiectasia refers to small dilated blood vessels, both of which are hallmarks of the disease. It affects many parts of the body; It impairs certain part of the brain including the cerebellum, causing difficulty with movement and coordination. It also weakens the immune system, causing a predisposition to infection and as well prevents repair of broken DNA, increasing the risk of cancer.

**Digeorge syndrome**: this is a chromosomal disorder caused by deletion of a small segment of chromosome 22. It results in poor development of severe bodily systems. Its features vary widely, even among members of the same family. It is typically due to the deletion of 30 to 40 genes in the middle of chromosome 22 at a location known as 22q11.2. About 90% cases occur due to a new mutation during early development, while 10% are inherited from a person’s parents. It is autosomal dominant, meaning that only one affected chromosome is needed for the condition to occur. Diagnosis is suspected based on the symptoms and confirmed by genetic testing.

**COMMON VARIABLE IMMUNODEFICIENCY**: it is one of the most frequently diagnosed primary Immunodeficiencies especially in adults, characterized by low levels of serum immunoglobulin and antibodies, which causes an increased susceptibility to infection. While CVID is thought to be due to genetic effects, the exact cause of the disorder is unknown. The degree and type of serum immunoglobulins and the clinical course, varies from patient to patient, hence the word variable. In some patients, there is a decrease in both IgG and IgA; in others, all three major types of immunoglobulins (IgA, IgG and IgM) are decreased. In others, there are defects of the T cells and this may also contribute to increased susceptibility to infections as well as autoimmunity, granulomata and tumors. To be sure that CVID is the correct diagnosis, there must be evidence of a lack of functional antibodies and other possible causes of these immunologic abnormalities must be excluded. Frequent and/or unusual infections may first occur during early childhood, adolescence or adult life. Patients with CVID have increased incidence of autoimmune or inflammatory manifestations, granulomata and an increased susceptibility to cancer when compared to the general population. Sometimes, it is presence of one of these other conditions that prompts an evaluation for CVID. The medical terms for absent or low blood immunoglobulins are **GAMMAGLOBULINEMIA AND HYPOGLOBULINEMIA** respectively. Due to the late onset of symptoms and diagnosis, other names that have been used in the past include; acquired globulinemia, adult onset gammaglobulinemia, or late onset hypogammaglobulinemia.

**SECONDARY IMMUNE DISODERS OR DISEASES:**  these are disorders that occur when the immune system is compromised especially due to an environmental factor and not genetic factor. It occurs when the immune system is weakened by another treatment or illness. Secondary immunodeficiency is also known as acquired deficiencies and it can result from various immunosuppressive agents. The secondary Immunodeficiency disorders include;

* **Cancer of the immune system (leukemia)**
* **HIV/Aids**

**CANCER OF THE IMMUNE SYSTEM (LEUKEMIA):** this 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. Symptoms include bleeding and bruising, feeling tired, fever, and an increased risk of infections. They occur due to lack of normal blood cells. The white blood cells crowd out the red blood cells and platelets that your body needs to be healthy. Many types exist such as acute lymphoblastic leukemia, acute myeloid leukemia and chronic lymphocytic leukemia. Leukemia symptoms may vary depending on the type of leukemia. Common leukemia signs and symptoms include;

* Fever or chills
* Bruising or bleeding easily
* Weakness of fatigue
* Infections that is severe or recurrent
* Headaches
* Pain in joints or bones
* Vomiting
* Seizures
* Weight loss
* Night sweats
* Shortness of breath
* Swollen lymph nodes

**HIV/AIDS:** HIV is a virus that damages the immune system. The immune system helps the body fight off infections. Untreated HIV infects and kills more CD4 cells, which are a type of immune cells called T cells. Over time the body is likely to get various types of infections and cancers. It is transmitted through bodily fluids (blood, semen, vaginal and rectal fluids, (breast milk). HIV is a lifelong condition and currently there is no cure, 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. It is caused by an RNA virus or retrovirus. HIV is transmitted to body fluids through blood, semen, vaginal and rectal fluids, and breast milk. The virus does not 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 is possible to manage HIV and live well with the virus for many years. Without treatment, a person is likely to develop a serious condition called AIDS. At that point, the immune system is too weak to fight off 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. If AIDS develops, it means that the immune system is severely compromised. It is weakened to the point where it can no longer fight off most diseases and infections. That makes the person vulnerable to a wide range of illnesses including;

* Pneumonia
* Tuberculosis
* Oral thrush; a fungal infection in the mouth or throat
* Cytomegalovirus (CMV), a type of herpes virus
* Cryptococcal meningitis; a fungal infection in the brain
* Toxoplasmosis; a brain infection caused by a parasite
* Cryptosporidiosis; an infection caused by an intestinal parasite
* Cancer; including Kaposi’s sarcoma and lymphoma