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QUESTIONS

Immunodeficiency disorder is the absence or failure of normal function of one or more elements of the immune system. There are two major types of immunodeficiency disorders: PRIMARY AND SECONDARY.

1. Identify and briefly explain 5 primary immunodeficiency disorders

2. Identify and briefly explain 2 secondary immunodeficiency disorders

ANSWER

Primary immunodeficiency

These are genetic origin and result from intrinsic defect in the cell. They represent inborn error of immune function that predispose people to infections. 5 primary immunodeficiency disorder are

1. Hyperimmunoglobulinemia E syndrome (HIE)

It is a type of phagocytic cell disorder. There is an increased incidence of bacterial and fungal infection caused by organism that are normally nonpathogenic. These patients may experience recurrent cutaneous abscesses, chronic eczema, bronchitis, pneumonia, chronic otitis media, and sinusitis. hyperimmunoglobulin E syndrome white blood cells cannot initiate an inflammatory response to infectious organisms. This results in recurrent bacterial infections of the skin and lung; abnormalities of connective tissue, skeleton, and dentition; and extremely elevated levels of IgA. patients with phagocytic cell disorders may be asymptomatic, severe neutropenia may present and may be accompanied by deep and painful mouth ulcers, gingivitis, stomatitis, and cellulitis. Death from overwhelming infection

Its treatment includes Antibiotic therapy and treatment for viral and fungal infections Granulocyte-macrophage colony stimulating

factor (GM-CSF); granulocyte colony-stimulating factor (G-CSF).

1. HYPOGAMMAGLOBULINEMIA syndrome (CVID)

This is a type of B-cell deficiency results from a lack of differentiation of B cells into plasma cells. plasma cells are the most vigorous producers of antibodies, affected patients have normal lymph follicles and many B lymphocytes that produce some antibodies. This syndrome is a frequently occurring immunodeficiency. It is also called CVID. Patients with CVID are susceptible to infections with encapsulated bacteria, such as Hemophilus influenzae.

 This disorder encompasses a variety of defects ranging from IgA deficiency, in which only the plasma cells that produce IgA are absent which result in severe panhypoglobulinemia (general lack of immunoglobulins in the blood). It usually presents within the first two decades of life, most patients are diagnosed as adults, because CVID often goes unrecognized. The etiology is still unknown but is believed to be multifactorial. Patients usually have normal B-cell lymphocyte counts, but the cells are clinically diverse and immature. They can recognize antigens due to their ability to become memory B cells

Treatment include Passive pooled plasma or gammaglobulin Intravenous immunoglobulin (IVIG), Metronidazole (Flagyl), Quinacrine HCl (Atabrine), Vitamin B12 and Antimicrobial therapy

1. DIGEORGE SYDROME/THYMIC HYPOPLASIA

This is a type of t- cell deficiency. This rare, complex, multisystem genetic abnormality, which affects multiple organ systems, has been mapped to chromosomes 10 or 22. The symptom variation is a result of differences in the amount of genetic material affected. T-cell deficiency occurs when the thymus gland fails to develop normally during embryogenesis. It is one of the few immunodeficiency disorders with symptoms that manifest almost immediately after birth.

 Infants born with DiGeorge syndrome have hypoparathyroidism

with resultant hypocalcemia resistant to standard therapy, congenital heart disease, cleft palate and lip, dysmorphic facial features, and possibly renal abnormalities. These infants are susceptible to yeast, fungal, protozoan, and viral infections and are particularly susceptible to

childhood diseases (chickenpox, measles, rubella), which are usually severe and may be fatal. The most frequent presenting sign in patients with DiGeorge syndrome is hypocalcemia that is resistant to standard therapy.

Treatment includes Thymus graft

1. SEVERE COMBINED IMMUNODEFICIENCY DISEASE(SCID)

This is a type of combined t-cell and b-cell deficiency. This is a disorder in which both B cells and T cells are missing. both cell-mediated and humoral functions are affected., SCID is marked by susceptibility to serious fungal, bacterial, and viral infections. It refers to a wide variety of congenital and hereditary immunologic defects characterized by early onset of infections, defects in both B-cell and T-cell systems, lymphoid aplasia, and thymic dysplasia. Inheritance of this disorder can be X linked, autosomal recessive, or sporadic.

The exact incidence of SCID is unknown; it is recognized as a rare disease in most population groups, with an incidence of about 1 case in 1,000,000. This illness occurs in all racial groups and both genders. Overwhelming severe fatal infections soon after birth (also includes opportunistic infections

 Treatment include Antimicrobial therapy; IVIG and bone marrow transplantation.

1. PAROXYSMAL NOCTURNAL HEMOGLOBINURIA (PNH)

This is a type of complement system. This is an acquired clonal stem cell disorder resulting from a somatic mutation in the hematopoietic stem cell. These include the complement-regulatory proteins the absence of which results in enhanced complement mediated lysis. Clinical manifestations may be indolent or life-threatening. The disorder is characterized by hemoglobinuria that increases during sleep, as well as intravascular haemolysis, cytopenia, infections, bone marrow hyperplasia, and a high incidence of life-threatening venous thrombosis, which occurs most commonly in the abdominal and cerebral veins.

 Severe fatigue, abdominal pain, and oesophageal spasm may also be present. Leukopenia, thrombocytopenia, and episodic crises are common. Severe infection can occur because of aplastic bone marrow and splenic thrombosis. Laboratory diagnosis can include specialized tests, such as the sucrose haemolysis test, Ham acid haemolysis test, and fluorescent activated cell analysis. A coagulation profile is also indicated

Secondary immunodeficiency

This is as result from external factors such as infection, the immune system can be affected by a variety of intrinsic factors, including immunosuppressive agents, harsh environmental conditions, hereditary disorders

1. HIV/AIDS

Acquired immunodeficiency syndrome (AIDS) is a chronic, potentially life-threatening condition caused by the human immunodeficiency virus (HIV). By damaging your immune system, HIV interferes with your body's ability to fight infection and disease.

HIV is a sexually transmitted infection (STI). It can also be spread by contact with infected blood or from mother to child during pregnancy, childbirth or breast-feeding. Without medication, it may take years before HIV weakens your immune system to the point that you have AIDS.

There's no cure for HIV/AIDS, but medications can dramatically slow the progression of the disease. These drugs have reduced AIDS deaths in many developed nations. HIV destroys CD4 T cells white blood cells that play a large role in helping your body fight disease. The fewer CD4 T cells you have, the weaker your immune system becomes. You can have an HIV infection, with few or no symptoms, for years before it turns into AIDS. AIDS is diagnosed when the CD4 T cell count falls below 200 or you have an AIDS-defining complication, such as a serious infection or cancer.

1. Cancer

 Cancer is a group of diseases involving abnormal cell growth with the potential to invade or spread to other parts of the body. These contrast with benign tumors, which do not spread. Possible signs and symptoms include a lump, abnormal bleeding, prolonged cough, unexplained weight loss, and a change in bowel movements. While these symptoms may indicate cancer, they can also have other causes. E.g. leukemia.

 When cancer begins, it produces no symptoms. Signs and symptoms appear as the mass grows or ulcerates. The findings that result depend on the cancer's type and location. Few symptoms are specific. Many frequently occur in individuals who have other conditions. Cancer can be difficult to diagnose and can be considered a "great imitate

 Cancer is fundamentally a disease of tissue growth regulation. For a normal cell to transform into a cancer cell, the genes that regulate cell growth and differentiation must be altered.

Treatment can be use of diet, radiation, medication to reduce its effects