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PRIMARY IMMUNODEFICIENCY DISORDERS

1) Thymus hypoplasia:

In thymic hypoplasia, thymic refers to the thymus which is an immune organ that sits between the lungs, hypo- refers to under, and -plasia refers to development.

Thymic hypoplasia is a condition where the thymus is underdeveloped and has a reduced number of cells leading to a reduced number of T-cells.

There are two main causes of thymic hypoplasia, DiGeorge syndrome, and Ataxia-telangiectasia syndrome, both cause a reduction of thymic cells.

In DiGeorge syndrome the parathyroid glands are underdeveloped, this results in less parathyroid hormones which results in hypocalcemia. In ataxia telangiectasia, there can also be symptoms like ataxia which is a problem with muscle coordination, telangiectasia, which are dilated blood vessels in the skin and eye, and an increased risk of cancer.

Diagnosing thymic hypoplasia often begins with genetic testing, which can help identify if it's DiGeorge syndrome or ataxia-telangiectasia syndrome.

And certain blood tests looking for T cell numbers and function can help determine the cause of recurrent infections.

Also, calcium levels in the blood may hint at the two conditions associated with thymic hypoplasia. In DiGeorge syndrome, there is hypocalcemia while in ataxia telangiectasia there's normal levels of calcium or eucalcemia.

Antibiotics may be used to treat infections from thymic hypoplasia.

Surgery might be required for more severe cases though, like a thymus transplant.

2) Immunoglobulin A deficiency

Selective immunoglobulin A deficiency (SIgAD) is a primary immunodeficiency disease and is the most common of the primary antibody deficiencies. Total immunoglobulin A deficiency (IgAD) is defined as an undetectable serum immunoglobulin A (IgA) level at a value < 5 mg/dL (0.05 g/L) in humans. Partial IgAD refers to detectable but decreased IgA levels that are more than 2 standard deviations below normal age-adjusted means.

IgAD is commonly associated with normal B lymphocytes in peripheral blood, normal CD4+ and CD8+ T cells, and, usually, normal neutrophil and lymphocyte counts. Anti-IgA autoantibodies of the IgG and/or IgE isotype may be present. Peripheral blood may also be affected by autoimmune cytopenias, eg, autoimmune thrombocytopenia, and patients may have other autoimmune phenomena.

3) Chronic Granulomatous Disease (CGD).

Occurs when white blood cells called phagocytes are unable to kill certain bacteria and fungi, making people highly susceptible to some 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.

4) 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.

5) Autoimmune Lymphoproliferative Syndrome (ALPS).

Autoimmune lymphoproliferative syndrome (ALPS) is a rare immune disorder first described by NIH scientists in the mid-1990s that can cause numerous autoimmune problems, such as low levels of red blood cells, clot-forming platelets, and infection-fighting white blood cells. These problems can increase the risk of infection and hemorrhage. Most cases of ALPS are caused by mutations in the FAS gene. FAS produces a receptor that, when activated, leads to programmed cell death, or apoptosis. This programmed death is an important part of the normal cell lifecycle. When cells do not receive the message that it is time for them to die, an abnormal buildup of cells can result.

SECONDARY IMMUNODEFICIENCY DISORDERS

1) LEUKEMIA

This is a cancer of the white blood cells(WBCs). WBCs are a vital part of your immune system. They protect your body from invasion by bacteria, viruses, and fungi, as well as from abnormal cells and other foreign substances. In leukemia, the WBCs do not function like normal WBCs. They can also divide too quickly and eventually crowd out normal cells. The types of leukemia are; (I) The onset of leukemia can be acute (sudden onset) or chronic (slow onset). In acute leukemia, cancer cells multiply quickly. In chronic leukemia, the disease progresses slowly and early symptoms may be very mild.

(II) Leukemia is also classified according to the type of cell. Leukemia involving myeloid cells is called myelogenous leukemia. Leukemia involving lymphocytes is called lymphocytic leukemia. There are four main types of leukemia:

i. Acute myelogenous leukemia (AML).
ii. Chronic myelogenous leukemia (CML).
iii. Acute lymphocytic leukemia (ALL).
iv. Chronic lymphocytic leukemia (CLL).

Symptoms of leukemia are;

i. Excessive sweating, especially at night.
ii. Fatigue and weakness.
iii. Weight loss.
iv. Bone pain and tenderness.

v. painless, swollen lymph nodes (especially in the neck and armpits).
vi. Enlargement of the liver or spleen.
vii. Red spots on the skin, called petechiae.
viii. Bleeding easily and bruising easily.
ix. Fever and chills.
x. Frequent infections.

2) **AIDS(Acquired Immunodeficiency Syndrome)**

This is the most advanced stage of HIV(Human Immunodeficiency Virus). HIV is a virus that damages the immune system.

H = Infects only Human beings

I = Immunodeficiency virus weakens the immune system and increases the risk of infection

V = Virus that attacks the body

Public awareness in 1981, First antiretroviral drug in 1987, PCP prevention started in 1988 (Pneumocystis pneumonia), 1995 protease inhibitors.

A = Acquired, not inherited

I = Weakens the Immune system

D = Creates a Deficiency of CD4+ cells in the immune systemS = Syndrome, or a group of illnesses taking place at the same time,

When the immune system becomes weakened by HIV, the illness progresses to AIDS

Some blood tests, symptoms or certain infections indicate progression of HIV to AIDS. African-Americans account for 50% of cases in 200, Kills more than 8000 cases per day worldwide,Now considered a disease of chronicity, Older adults at risk for HIV infection, Women are fastest growing group w/HIV, Women have poorer outcomes , Fatality rate is 60%, Hits hardest between 25-44 years of age.

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 has no cure. However, with medical care, including treatment called antiretroviral therapy, it's possible to manage HIV and live with the virus for many years. HIV is transmitted through bodily fluids that include: blood, semen, vaginal and rectal fluids, breast milk. 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. The life expectancy of someone with AIDS is 3 years.

REFERENCE
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