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TITLE: IMMUNODEFICIENCY DISORDERS

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LEVEL: 300

IMMUNODEFICIENCY DISORDERS ARE:

PRIMARY IMMUNODEFICIENCY DISORDERS

1. Autoimmune lymphoproliferative syndrome (ALPS).

2. Chronic Granulomatous Disease (CGD)

3. Severe Combined Immunodeficiency (SCID)

4. Congenital Neutropenia syndromes

5. Leukocyte adhesion deficiency (LAD)

(1). AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME(ALPS): This 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 life cycle. When cells do not receive the message that it is time for them to die, an abnormal buildup of the cells can result.

(2). CHRONIC GRANULOMATOUS DISEASE (CGD): This occurs when white blood cells called phagocytes are unable to kill certain Bacteria and fungi, making people highly susceptible to some bacteria and fungi infections. It is an inherited primary immunodeficiency disease which increases the body’s susceptibility to infections caused by certain bacteria and fungi. Granulomas are masses of immune cells that form at sites of infection or inflammation. People with this condition are unable to fight off common germs and get very sick from infection that will be mild in healthy people. This is because the disorder makes it difficult for neutrophils to produce hydrogen peroxide. These sever infections can include skin or bone infections and abscesses in internal organs such as lungs, liver or brain.

CAUSES: A mutation in one of five genes can cause chronic granulomatous disease. A mutated gene can be inherited from a parent.

RISK FACTORS: Gender; male children are more likely to have it.

SIGNS AND SYMPTOMS: children with CGD develop severe infections in infancy or early childhood, development of a serious type of fungal pneumonia after being exposed to dead leaves, mulch or hay.

TREATMENT: Therapeutic management for CGD includes prophylactic antibiotics and antifungal medications, interferon-gamma injections and aggressive management of acute infections. Bone marrow transplant can cure chronic granulomatous disease.

(3). SEVERE COMBINED IMMUNODEFICIENCY: It is a group of rare, life-threatening disorders that typically presents in infancy and 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 primary immunodeficiency disease. It is a genetic defect that affects the function of the T cells.

PROGNOSIS: Affected infants will often die within the first year of life without hematopoietic stem cell transplant.

CAUSES: It is caused by mutation in different genes involved in development and function of B and T cells.

TYPES: The most common type is linked to a defect in a gene on the X chromosome, affecting only males. Other forms are caused by a deficiency of adenosine deaminase and a variety of other genetic defects.

SIGNS AND SYMPTOMS: Frequent and often very severe respiratory infections, poor growth, eczema looking rashes, chronic diarrhea, recurrent thrush in mouth, pneumocystis pneumonia.

TREATMENT: The only treatment available presently is bone marrow transplant. The earlier severe combined immunodeficiency is detected, the higher the success of the treatment.

(4). CONGENITAL NEUTROPENIA SYNDROME: Congenital neutropenia syndromes are a group of disorders present from birth that are characterized by low levels of neutrophils, a type of white blood cell necessary for fighting infections. They are a group of rare disorders present from birth that are characterized by low levels of neutrophils. It may also be referred to as congenital agranulocytosis, severe congenital neutropenia, severe infantile genetic neutropenia, infantile genetic agranulocytosis, or Kostmann disease. It manifests in infancy by age 3 months with life-threatening bacterial infections. The mouth and perirectum are the most common sites of infection. It is mostly found in remote, isolated populations with a high degree of consanguinity.

CAUSES: It is caused by genetic mutation of any of the following genes; ELANE, HAX1, G6PC3, GFI1, CSF3R, X-linked WAS, CXCR4, VPS45A, and JAGN1.

SIGNS AND SYMPTOMS: Fever, pneumonia, gingivitis, stomatitis, periodontitis, recurrent oral ulcerations, premature loss of teeth. People with congenital neutropenia experience bacterial infections early in like. These may cause inflammation of the umbilical cord stump, abscesses (or boils) on the skin, oral infections and pneumonia.

TREATMENT: The standard therapy for congenital neutropenia includes injections of granulocyte colony-stimulating factor (G-CSF), which can help restore immune system function. Bone marrow transplant may also be done.

(5). LEUKOCYTE ADHESION DEFICIENCY (LAD): It is a rare autosomal recessive disorder characterized by immunodeficiency resulting in recurrent infections. It is a defect of cellular adhesion molecules resulting in clinical syndromes. It is a combined B and T cell disorder. Its major immunodeficiency features are:

* Inability to form pus
* Deficiency of various glycoproteins including LFA-1/Mac-1, glycoprotein 50/95
* Inability of leukocytes to migrate to infection sites to kill invading microorganisms due to mutations in the CD18 glycoprotein.
* Adhesion molecules deficiency results in abnormal inflammatory response and eventually recurrent bacterial infections.

TYPES: LAD1, LAD2, LAD3

CAUSES: Genetic mutation of the ITGB2 gene.

SIGNS AND SYMPTOMS: Recurrent bacterial infections, defects in neutrophil adhesions, delay in umbilical cord sloughing, inability to form pus, bacterial infections such as omphalitis, pneumonia, gingivitis, peritonitis are common.

TREATMENT: The only current curative therapy is the hematopoietic stem cell transplant, although patients can also receive intensive antibiotherapy and granulocyte transfusions from healthy donors.

COMPLICATIONS: Infectious diseases affecting the skin, respiratory system, gastrointestinal system, oral cavity and some internal organs.

SECONDARY IMMUNODEFICIENCY DISORDERS

1. Chronic leukemia

2. Human immunodeficiency Virus (HIV)

1. CHRONIC LEUKEMIA: It is a cancer of blood-forming tissues hindering the body’s ability to fight infection. This is cancer of the WBCs. WBCS are a vital part of the immune system which protect our body from invasions by bacteria, viruses and fungi as well as normal WBCs. It is caused by rise in the number of white blood cells in the body. These white blood cells crowd out the red blood cells and platelets that the body needs to be healthy. The additional white blood cells (leukemia cells) do not fight infections.

Types of leukemia are:

1. 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.

2. Leukemia can also be classified according to the type of cell. Leukemia involving myeloid cells is called myelogenous leukemia. Leukemia involving lymphocytes is called lymphocytes leukemia. There are four main type of leukemia:

. Acute myelogenous leukemia (AML).

.Chronic myelogenous leukemia (CML).

.Acute lymphocytes leukemia (ALL).

.Chronic lymphocytes leukemia (CML).

RISK FACTORS: Smoking, exposure to radiation and certain chemicals, people that received radiation therapy or chemotherapy, family history of leukemia, genetic disorders like Down syndrome.

CAUSES: The exact cause is not known.

SIGNS AND SYMPTOMS: Pain in the bones and joints, dizziness, fatigue, fever, loss of appetite, bleeding, easy bruising, mouth ulcers, nosebleed, pale skin, petechiae, dyspnea, swollen lymph nodes, weight loss, weakness, recurrent infections, headaches, seizures.

TREATMENT: Chemotherapy, radiation theraphy, biologic therapy, hematopoietic stem cell transplant, surgery (splenectomy), medications (Imatinib).

2. HUMAN IMMUNODEFICIENCY VIRUS (HIV): It is a sexually transmitted infection which interferes with the body’s ability to fight infection and disease by damaging the immune system. It can develop into AIDS (Acquired immunodeficiency syndrome) if left unmanaged. There is no cure for it but it can be managed.

CAUSE: It is caused by the human immunodeficiency virus.

RISK FACTORS: People who have unprotected sex, those who have STIs, IV drug users, homosexuals.

MODES OF TRANSMISSION: It can be transmitted through infected body fluids (blood, seminal fluids, vaginal secretions, amniotic fluid, and breast milk), from mother to child during pregnancy, child birth, or breast feeding, transfusion of infected blood and blood products, sharing of needles.

STAGES: HIV if left untreated has four stages and they are:

* INFECTION: This is the first stage. In this stage, HIV quickly replicates in the body after infection. Some people develop short-lived flu-like symptoms and a rash within days to weeks after infection. During this time, the immune system reacts to the virus by developing antibodies (sero-conversion).
* ASYMPTOMATIC STAGE: This stage does not show any outward signs or symptoms. The virus continues to weaken the immune system even though the person looks well physically. This stage may last for years.
* SYMPTOMATIC STAGE: The immune system has been damaged and weakened by HIV and symptoms develop. Initially they may be mild but they do worsen. The symptoms are caused by the emergence of opportunistic infections such as tuberculosis, toxoplasmosis e.t.c.
* PROGRESSION TO AIDS: The virus develops into full blown AIDS. This stage is not inevitable.

SIGNS AND SYMPTOMS: It depends on the stage of the infection.

* INFECTION STAGE: Signs and symptoms include fever, headache, muscle aches and joint pain, rash, sore throat, diarrhea, weight loss, cough, night sweats.
* SYMPTOMATIC STAGE: Fever, fatigue, swollen lymph nodes, diarrhea, weight loss, thrush, shingles, pneumonia.
* PROGRESSION TO AIDS: Sweats, chills, recurring fever, chronic diarrhea, persistent lesions in the mouth, persistent unexplained fatigue, weakness, weight loss, skin rashes and bumps.

TREATMENT: It can be managed through the use of antiretroviral drugs.

PREVENTION: Preventions include:

* Use of the medication as prevention so that your partner does not get it if you are already infected.
* Use post-exposure prophylaxis if you’ve been exposed to HIV.
* Use a new condom every time you have sex
* Use a clean needle
* Do not have unprotected sex

COMPLICATIONS: Some complications of HIV are: thrush, tuberculosis, cytomegalovirus, toxoplasmosis, lymphoma, wasting syndrome e.t.c.