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MED SURG' ASSIGNMENT
300L

1. Primary immunodeficiency disorders

A group of genetically determined disorders characterized by an impaired ability to produce normal immune response caused by mutations in genes involved in the development and function of immune organs, cells and molecules.

Examples include;

I. Severe combined immunodeficiency(SCID)
II. DiGeorge's syndrome
III. Ataxia-telangiectasia
IV. Chediak-Higashi syndrome
V. Leucocyte- Adhesion defect

I. Severe combined immunodeficiency(SCID)

Severe combined immunodeficiency (SCID) is a group of rare disorders caused by mutations in different genes involved in the development and function of infection-fighting immune cells. Infants with SCID appear healthy at birth but are highly susceptible to severe infections.

It is characterized by;

Absence of T-lymphocytes,

Impairment of both cellular and humoral response,

Specific defects in antigen presentation and functional immune molecules.

Features include;

- Absent tonsils
- Small or absent lymph nodes
- Absent thymic shadow
- Lymphopenias
- Decreased number of T cells
- Severe agammaglobulinemia
- No IG's usually present

II. DiGeorge's syndrome

Also called thymic hyperplasia

DiGeorge Syndrome (DGS) is a primary immunodeficiency, often but not always, characterized by cellular (T-cell) deficiency, characteristic facies,

congenital heart disease and hypocalcemia. DGS is caused by abnormal formation of certain tissues during fetal development.
Approximately 90% of patients with DGS have a small deletion in chromosome number 22 at position 22q11.2.
Features include;

- Heart defects
- Thymus gland abnormalities
- Cleft palate
- Antimongoloid eyes
- Short philtrum with fish-mouth appearance
- Micrognathia
- Low set pixie-like ears
- Short palpebral fissures

III. Ataxia-telengiectasia

Ataxia-telangiectasia is a rare inherited disorder that affects the nervous system, immune system, and other body systems. This disorder is characterized by progressive difficulty with coordinating movements (ataxia) beginning in early childhood, usually before age 5. Affected children typically develop difficulty walking, problems with balance and hand coordination, involuntary jerking movements (chorea), muscle twitches (myoclonus), and disturbances in nerve function (neuropathy)
Mutations in the ATM gene cause ataxia-telangiectasia. The ATM gene provides instructions for making a protein that helps control cell division and is involved in DNA repair.
Characteristics- Progressive cerebellar ataxia
Ocular and cutaneous telangiectasia
Severe sino-pulmonary infections
Progeric changes
Sclerodermoid changes

IV. Chediak-Higashi syndrome

Chediak-Higashi syndrome (CHS) is a rare, inherited, complex, immune disorder that usually occurs in childhood characterized by reduced pigment in the skin and eyes (oculocutaneous albinism), immune deficiency with an increased susceptibility to infections, and a tendency to bruise and bleed easily. Neurological deficits are also common. CHS is transmitted as an autosomal recessive genetic condition.

Chediak-Higashi syndrome is inherited as an autosomal recessive genetic trait. The responsible gene has been mapped to chromosomal locus 1q42.1-q42.2 and is known as LYST gene.
Abnormal granules in leucocytes leading to

- Hypopigmentation/partial albinism
- Severe immunodeficiency
- Neurologic abnormalities
- Mild bleeding tendencies

V. Leucocyte adhesion defect

Leukocyte adhesion deficiency (LAD) is a rare primary immunodeficiency. The clinical picture is characterized by marked leukocytosis and localized bacterial infections that are difficult to detect until they have progressed to an extensive level secondary to lack of leukocyte recruitment at the site of infection.
Theres a defect in CD18 gene
Characteristics; Delayed cord dehiscence and scar formation
Recurrent diarrhea and respiratory symptoms
Leucocytosis

2. Secondary immunodeficiency disorders

Secondary immunodeficiencies result from the adverse consequences of exposure to a variety of factors including infectious agents, drugs, metabolic diseases, and environmental conditions
These conditions may affect the immune function in a manner that varies in presentation and severity, and the restoration of complete immunity is achieved with the improvement of the primary disease or the removal of the offending agent.
Examples include;

I. Human immunodeficiency virus (HIV) infection
II. Diabetes

I. Human immunodeficiency virus (HIV) infection

Human immunodeficiency virus (HIV) infection is a viral infection that progressively destroys certain white blood cells and can cause acquired immunodeficiency syndrome (AIDS).
HIV progressively destroys certain types of white blood cells called CD4+ lymphocytes. Lymphocytes help defend the body against foreign cells, infectious organisms, and cancer. Thus, when HIV destroys CD4+

lymphocytes, people become susceptible to attack by many other infectious organisms. Many of the complications of HIV infection, including death, usually result from these other infections and not from HIV infection directly.
HIV is transmitted through close contact with a body fluid that contains the virus or cells infected with the virus (such as blood, semen, or vaginal fluids).
HIV destroys certain types of white blood cells, weakening the body's defenses against infections and cancers.
When people are first infected, symptoms of fever, rashes, swollen lymph nodes, and fatigue may last a few days to several weeks.
Many infected people remain well for more than a decade.
About half of untreated people become ill and develop AIDS, defined by the presence of serious infections and cancers, within about 10 years.
Eventually, most untreated people develop AIDS.
Blood tests to check for HIV antibody and to measure the amount of HIV virus can confirm the diagnosis.
HIV drugs (antiretroviral drugs)—two, three, or more taken together—can stop HIV from reproducing, strengthen the immune system, and thus make people less susceptible to infection, but the drugs cannot eliminate HIV, which persists in an inactive form.

II. Diabetes mellitus

Diabetes mellitus, commonly known as diabetes, is a group of metabolic disease that causes high blood sugar. The hormone insulin moves sugar from the blood into your cells to be stored or used for energy. With diabetes, your body either doesn't make enough insulin or can't effectively use the insulin it does make.
Untreated high blood sugar from diabetes can damage your nerves, eyes, kidneys, and other organs.

There are a few different types of diabetes:

- Type 1 diabetes is an autoimmune disease. The immune system attacks and destroys cells in the pancreas, where insulin is made. It's unclear what causes this attack. About 10 percent of people with diabetes have this type.
- Type 2 diabetes occurs when your body becomes resistant to insulin, and sugar builds up in your blood.
- Gestational diabetes is high blood sugar during pregnancy. Insulin-blocking hormones produced by the placenta cause this type of diabetes.