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Course ; medical surgical nursing

Answers

1. **Primary immunodeficiency disorders are;**
* Chronic granulomatous disease (CGD)
* Congenital neutropenia syndromes
* CTLA4 deficiency
* DOCK8 deficiency
* Hyper –immunoglobulin E syndromes (HIES)
1. **Chronic granulomatous disease (CGD)**

Chronic granulomatous disease is a genetic disorder in which white blood cells called phagocytes are unable to kill certain types of bacteria and fungi.

People with CGD are highly susceptible to frequent life- threatening bacterial and fungal infections.

**Causes;** it is causedby 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.

**Signs and symptoms;** people with CGD are highly susceptible to infections caused by certain bacteria and fungi, such as staphylococcus aureus, Serratia marcescens, and Aspergillus species. These people may develop abscesses (boils) in their lungs, liver, spleen, bones, or skin; and masses of cells, called granulomatous, that can obstruct the bowel or urinary tract.

**Treatment;** people with CGD take lifelong regimes of antibiotics and antifungals to prevent infections. Granulomatous may require steroid therapy. Some people with CGD have been treated successfully with **bone marrow transplantation** .

1. **Congenital neutropenia syndromes**

Congenital neutropenia syndromes are a group of rare disorders present from birth that are characterized by low levels of **neutrophils**, a typeof whiteblood cell necessary for fighting infections. Congenital neutropenia syndromes also may be referred to as congenital agranulocytosis, severe congenital neutropenia, severe infantile genetic neutropenia, infantile genetic agranulocytosis, or kostmann disease.

**Causes;** Researchers have identified numerous genetic mutations that cause congenital neutropenia syndromes. Generally, mutations that result in congenital neutropenia affect the development, lifespan or function of neutrophils. Congenital neutropenia syndromes are inherited through autosomal recessive, autosomal dominant and X-linked inheritance patterns. The genes linked to this syndromes includes the following;

* ELANE
* HAX1
* G6PC3
* GFI1
* X-linked WAS
* JAGN1

**Treatment;** Standard therapy for congenital neutropenia includes injections of granulocyte colony-stimulating factor (G-CSF), which can help restore immune system function.

1. **CTLA4 deficiency;** CTLA4 deficiency is a rare disorder that severely impairs the normal regulation of the immune system, resulting in conditions such as intestinal disease, respiratory infections, autoimmune problems, and enlarged lymph nodes, liver and spleen.

**Signs and Symptoms;** CTLA4 is characterized by infiltration of immune cells into the gut, lungs, bone marrow, Central nervous system, kidneys, and possibly other organs. Most people with CTLA4 deficiency experience diarrhea or intestinal disease, enlarged lymph nodes, liver and spleen also are common as respiratory infections.

**Treatment;** Treatment for CTLA4 deficiency may include standard therapies for auto immune problems and immunoglobulin deficiencies.

1. **DOCK8 deficiency**; DOCK8 deficiency is a rare immune disorder named after the mutated gene responsible for the disease. DOCK8 deficiency is associated with very high levels of an anti-body called immunoglobulin E, or igE.

**Signs and Symptoms**; DOCK8 immunodeficiency syndrome is characterized by recurrent infections, allergies and certain cancers. They may also experience recurrent chronic upper and lower respiratory tract infection.

**Treatment**; Doctors may recommend the prophylactic, or preventive, use of anti- microbial drugs to prevent infections, they may also consider using immunoglobulin replacement therapy.

1. **Hyper-immunoglobulin E syndromes (HIES);** Many different syndromes are known to lead to high levels of an antibody called immunoglobulin E , or igE. Collectively, these conditions are called hyper-igE syndromes, or HIES. Other conditions, such as severe eczema, can lead to extremely high igE levels that are not caused by a syndrome at all.

Each set of mutations causes a specific syndrome, including autosomal- dominant CARD11 deficiency, DOCK8 deficiency, IL6R deficiency, IL6ST deficiency, PGM3 deficiency, STAT3 dominant- negative disease and many more others.

1. **Secondary immunodeficiency disorders are**;
* HIV /AIDS
* Leukemia
1. **HIV /AIDS**; AIDS is a disease that can develop in people with HIV. It’s the most advanced stage of HIV. But just because a person has HIV doesn’t mean they will develop AIDS.

HIV kills CD4 cells. Healthy adults generally have a CD4 count of 500 to 1500 per cubic millimeter. A person can also be diagnosed with HIV. A person can also be diagnosed with AIDS if they have HIV and develop an opportunistic infection, such as pneumonia , one that takes advantage of a unique situation, such as HIV.

Untreated HIV can progress to AIDS within a decade. There is no cure for AIDS, and without treatment, life expectancy after diagnosis is about three years. This may be shorter if the person develops a severe opportunistic illness. However , treatment with antiretroviral drugs can prevent AIDS from developing.

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. That makes the person vulnerable to a wide range of illnesses, including;

* Pneumonia
* Tuberculosis

HIV can be transmitted through bodily fluids which include;

* Blood
* Semen
* Vaginal and rectal fluids
* Breast milk

**Symptoms of HIV include**;

* Fever
* Chills
* Swollen lymph nodes
* General aches and pains
* Skin rash
* Weight loss

 **Symptoms of AIDS include;**

* Recurrent fever
* Chronic swollen lymph glands, especially around the armpits, neck, and groin
* Chronic fatigue
* Night sweats
* Dark splotches under the skin or inside the mouth, nose, or eyelids.
1. **Leukemia;** This is a blood cancer caused by a rise in the number of white blood cells in the body.

Those white blood cells crowd out the red blood cells and platelets that the body needs to be healthy. The extra white blood cells don’t work right.

There are four main types of leukemia which include;

* Acute lymphocytic leukemia (ALL)
* Acute myelogenous leukemia (AML)
* Chronic lymphocytic leukemia(CLL)
* Chronic myelogenous leukemia (CML)

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**Signs and symptoms;**

* Weakness or fatigue
* Bruising or bleeding
* Infections that are severe or keep coming back
* Pain in bones or joints
* Headaches
* Vomiting
* Seizures
* Weight loss
* Night sweats
* Shortness of breath
* Swollen lymph nodes or organs like spleen