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Course; med surge

NSC 306

Five forms of primary disorder;

1. 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 haemorrhage.

Causes

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 build-up of cells can result. In the case of ALPS, mutations in FAS cause an abnormal build-up of white blood cells.

2. APS-1 (APECED)

Autoimmune polyglandular syndrome type 1 (APS-1), also called autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED), causes a diverse range of symptoms, including autoimmunity against different types of organs and increased susceptibility to candidiasis, a fungal infection caused by Candida yeast.

The syndrome is caused by mutations in the autoimmune regulator (AIRE) gene. NIAID researchers are exploring how AIRE mutations impact the function of cells by studying people with APS-1 (APECED), as well as using mouse models of AIRE deficiency. With a better understanding of how these mutations affect the immune system and other cells in the body, researchers hope to uncover promising therapeutic targets to prevent and treat persistent candidiasis and autoimmune disorders.

3. BENTA disease

BENTA disease is a rare genetic disorder of the immune system caused by mutations in the gene CARD11. The disease is characterized by high levels of certain immune cells starting in infancy, an enlarged spleen, enlarged lymph nodes, immunodeficiency, and an elevated risk of lymphoma, a type of cancer

BENTA disease is inherited in an autosomal dominant manner. “Autosomal” refers to the fact that every person has two copies of the CARD11 gene, one inherited from each parent. Only one of the two copies of CARD11 needs to be abnormal for a person to have BENTA disease. “Dominant” indicates that the abnormal copy of the gene from one parent dominates the matching, normal gene copy from the other parent. Autosomal dominant inheritance means that most families with BENTA disease have affected relatives on the side of the family with the mutation.

Children of a parent who carries a CARD11 mutation have a 50 percent chance of inheriting the mutation. In a family, each child’s risk of inheriting a mutated copy of the CARD11 gene is independent of whether his or her siblings inherited the mutation. For example, if the first four children in a family have the mutation, the next child has the same 50 percent risk of inheriting the mutation. Children who do not inherit the mutation will not develop BENTA disease or pass on the mutation to their children.

BENTA disease can also arise spontaneously in a patient as the result of a de novo mutation in CARD11. De novo mutations are not inherited but occur as a result of a mutation in the egg or sperm of one of the parents or in the fertilized egg itself. A patient with a de novo mutation can pass on the mutation to his or her children

4. Common variable immunodeficiency (CVID)

CVID is caused by a variety of different genetic abnormalities that result in a defect in the capability of immune cells to produce normal amounts of protective antibodies. People with CVID experience frequent bacterial and viral infections of the upper airway, sinuses, and lungs

Causes

CVID is caused by a variety of different genetic abnormalities that result in a defect in the capability of immune cells to produce normal amounts of all types of antibodies. Only a few of these defects have been identified, and the cause of most cases of CVID is unknown. Many people with CVID carry a DNA variation called a polymorphism in a gene known as TACI. However, while this genetic abnormality confers increased risk of developing CVID, it alone is not capable of causing CVID.

CVID is also linked to IgA deficiency, a related condition in which only the level of the antibody immunoglobulin A (IgA) is low, while levels of other antibody types are usually normal or near normal. IgA deficiency typically occurs alone, but in some cases it may precede the development of CVID or occur in family members of CVID patients.

5. Chronic granulomatous disease (CGD)

Chronic granulomatous disease (CGD) 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 and sometimes life-threatening bacterial and fungal infections.

Causes

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.

2. Two secondary immunodeficiency syndrome;

1. Down syndrome;

Down syndrome is a genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. This extra genetic material causes the developmental changes and physical features of Down syndrome.

Down syndrome varies in severity among individuals, causing lifelong intellectual disability and developmental delays. It's the most common genetic chromosomal disorder and cause of learning disabilities in children. It also commonly causes other medical abnormalities, including heart and gastrointestinal disorders.

Causes

Human cells normally contain 23 pairs of chromosomes. One chromosome in each pair comes from your father, the other from your mother.

Down syndrome results when abnormal cell division involving chromosome 21 occurs. These cell division abnormalities result in an extra partial or full chromosome 21. This extra genetic material is responsible for the characteristic features and developmental problems of Down syndrome. Any one of three genetic variations can cause Down syndrome:

• Trisomy 21. About 95 percent of the time, Down syndrome is caused by trisomy 21 — the person has three copies of chromosome 21, instead of the usual two copies, in all cells. This is caused by abnormal cell division during the development of the sperm cell or the egg cell.

• Mosaic Down syndrome. In this rare form of Down syndrome, a person has only some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.

• Translocation Down syndrome. Down syndrome can also occur when a portion of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. These children have the usual two copies of chromosome 21, but they also have additional genetic material from chromosome 21 attached to another chromosome.

2. 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’ll develop AIDS.

HIV kills CD4 cells. Healthy adults generally have a CD4 count of 500 to 1,500 per cubic millimetre. A person with HIV whose CD4 count falls below 200 per cubic millimetre will be diagnosed with AIDS.

A person can also be diagnosed with AIDS if they have HIV and develop an opportunistic infection or cancer that’s rare in people who don’t have HIV. An opportunistic infection, such as pneumonia, is one that takes advantage of a unique situation, such as HIV.

Untreated, HIV can progress to AIDS within a decade. There’s no cure for AIDS, and without treatment, life expectancy after diagnosis is about three years

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. 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

• Oral thrush, a fungal infection in the mouth or throat

• Cytomegalovirus (CMV), a type of herpes virus

• Cryptococci meningitis, a fungal infection in the brain

• Toxoplasmosis, a brain infection caused by a parasite

• Cryptosporidiosis, an infection caused by an intestinal parasite

• Cancer, including Kaposi’s sarcoma (KS) and lymphoma

The shortened life expectancy linked with untreated AIDS isn’t a direct result of the syndrome itself. Rather, it’s a result of the diseases and complications that arise from having an immune system weakened by AIDS.