UNIT 2

DISEASE GUIDE

PRIMARY IMMUNODEFICIENCIES

What are primary immunodeficiencies?

Primary immunodeficiencies is the name given to a group of more than 200 hereditary diseases caused by defects in the genes of the cells that make up the immune system.

As a result, the immune system does not carry out its function of protecting the body from the infectious action of microorganisms such as bacteria or viruses. The weakness of the immune system by a gene defect that prevents proper operation is the cause of the so-called primary or congenital immunodeficiency.

The signs of immunodeficiency can occur at all stages of life: childhood, adolescence or adulthood.

Symptoms

Infection susceptibility is one of the most common symptoms. The infections may occur in the skin, the sinuses, the throat, the ears, the lungs, the brain, the spinal cord, and/or the urinary or intestinal tracts. The increased vulnerability to infection may include repeated infections, infections that won't clear up, or unusually severe infections. People with primary immunodeficiency live their entire lives being more susceptible to infections, enduring recurrent health problems and often developing serious and debilitating illnesses. Fortunately, with proper medical care, many patients live full and independent lives.

- Severe infection – requires hospitalization or intravenous antibiotics
- Persistent infection – won't completely clear up or clears up very slowly
- Unusual infection – caused by an uncommon organism
- Recurrent infection – keeps coming back
- Runs in the family – others in the family have had a similar issue

Treatment

Primary immunodeficiency disease (PIDD) is treated with immunoglobulin preparations, which are plasma proteins. Immunoglobulins can be administered intravenously, which means they are injected into the vein, or subcutaneously as well for PIDD. The doctor determines the dose according to the patient's weight and physical condition. This medicine can be administered in a day unit at the hospital or in infusion centers. The full treatment usually takes about 4 hours.

While this medication, which is extracted from plasma, replaces the antibodies that the body doesn't produce, it doesn't help a weak immune system to create its own immunoglobulins. Therefore, in order to ensure the patient's quality of life, these substitution treatments must be taken regularly.

UNIT 2

DISEASE GUIDE

HEMOPHILIA

What is hemophilia?

Hemophilia is a recessive genetic disease, thus inherited, which may not manifest in the person who has it, but may appear in their children or grandchildren. This condition hinders good blood coagulation due to the deficiency of a blood coagulation factor.

Coagulation factors are plasma components involved in bleeding control. The body's inability to form the VIII and IX coagulation factors is associated with a defective gene found on the X chromosome.

The X chromosome is one of the two genes that determine a person's gender, that's why we say that it is hereditary.

There are two main types of hemophilia, depending on the clotting factor, in which a decrease or absence is observed:

- Hemophilia A: absence or deficiency of the VIII factor.
- Hemophilia B: absence or deficiency of the IX factor.

This is a rare disease and is less common in women. Of approximately 10,000 people, 1 is born with hemophilia.

Symptoms

In normal clotting, blood is always within the blood vessels, so they block the blood's exit to the tissues that surround it. The usual manifestation of hemophilia is internal bleeding.

- Hemarthrosis: internal bleeding of a joint, for example, a knee. They are the most common and are characterized by pain, an increasing size of the joint and loss of motion.
- Muscle hematomas: they occur spontaneously, either by trauma or sprains. They are characterized by pain, increased volume, local heat, and loss of function.

Treatment

Hemophilia is treated by replacing the VIII or IX factor to maintain stable protein levels in the bloodstream or when bleeding occurs. These factors, present in the plasma of healthy persons, provide the proteins that the patient's blood lacks for proper blood clotting.

UNIT 2

DISEASE GUIDE

ALPHA-1 ANTITRYPSIN DEFICIENCY

What is alpha-1 antitrypsin deficiency?

Alpha-1 antitrypsin deficiency, also known as alpha-1, is a condition people are born with. Alpha-1 can typically present as liver and/or lung disease. Alpha-1 antitrypsin (AAT) is a protein primarily produced in the liver and a lesser amount in the lungs. One of the major functions of the protein is to protect the lung from damage due to infections, stress, and toxic exposures, e.g. inhaled chemicals/particles, tobacco smoke.

In typical individuals AAT protein made in the liver is released into the blood. The proteins are made from normal genes, which are an inherited component of every cell that direct specific biological functions.

In affected individuals, altered genes make abnormally shaped AAT proteins. The abnormal protein is retained in the liver and cannot be fully released into the blood, creating a deficiency in the body.

People with alpha-1 antitrypsin deficiency most commonly develop liver disease because the abnormal AAT protein gets stuck in the liver. They can also develop lung disease because less normal AAT protein is able to get to the lungs to protect them. Severe alpha-1 antitrypsin deficiency may eventually cause chronic obstructive pulmonary disease (COPD), and do so at a relatively young age (30 to 40 years old).

Signs and symptoms

There are many different symptoms that can go along with Alpha-1. Each person with Alpha-1 may have different symptoms, and it is possible that some people won't get sick at all. Some of those symptoms are:

- Trouble breathing
- Wheezing
- Coughing a lot
- Unexplained liver disease
- Bronchiectasis, an illness that causes the tubes leading to the lungs to be permanently widened. It can lead to infections
- Recurrent chest colds
- Coughing up blood
- Jaundice (This is when the eyes and skin have a slightly yellow color)
- Panniculitis (Tender skin nodules)

Treatment

The diagnosis of alpha-1 is usually determined by three rather sophisticated laboratory tests that require a small sample of blood.

Adults with lung disease due to AAT deficiency may be treated with augmentation therapy intravenously.This treatment uses a needle to place the alpha-1 protein directly back into the body. This helps increase the levels of protein in the blood. The AAT protein is purified from the plasma of human donors. Augmentation therapy is not for children and is not a treatment for alpha-1 related liver disease.