EUROPEAN JOURNAL OF LIFE SAFETY AND STABILITY (EJLSS)
ISSN2660-9630
www.ejlss.indexedresearch.org Volume 27, March-2023 ||

Immunopathological Processes, Autoimmune Diseases, Bruton's Disease, Pathology of Di Giorgi Syndrome

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Abstract: *The field of medicine is an integral part of every society and society. It is not a secret to anyone that major reforms are being carried out to develop this sector, including in our country. In this article Immunopathological processes. Autoimmune diseases. Bruton's disease. Pathology of Di Giorgi syndrome is described in detail.*

Keywords: *pathology, medicine, Di Giorgi syndrome, diseases, new method of treatment, etc.*

Date of Submission: 11-02-2023 ***Date of Acceptance: 13-03-2023***

The DiGeorge syndrome is a pathology of genetic origin manifested by the development of malformations related to the structure of the heart, face, thymus and parathyroid glands. At the clinical level, they cause a variety of medical complications, including immunodeficiency, hypocalcemia, cardiac disease, and psychiatric disorders. As for the etiological origin, it is related to the genetic change of the 22nd chromosome. For this reason, it is also called 22q11.2 deletion syndrome. The diagnosis is based on the determination of cardinal clinical signs through physical examination and various laboratory tests: analytical and immunological examination, abdominal ultrasound, echocardiogram and genetic studies, mainly based on in situ hybridization (FISH). DiGeorge syndrome is associated with hypo- or aplasia of the thymus and parathyroid glands, leading to T-cell immunodeficiency and hypoparathyroidism. Infants with DiGeorge syndrome have low ear position, midline facial cleft, mild mandibular underdevelopment, hypertelorism, short philtrum, developmental delay, and congenital heart disease. Diagnosis is based on clinical findings and includes assessment of immune and parathyroid function, and chromosomal analysis. Treatment includes supportive care and, in severe cases, thymus or stem cell transplantation. DiGeorge syndrome is a primary immunodeficiency that includes T-cell defects. This is the result of deletions at the DiGeorge 22q11 chromosomal locus on chromosome 22, gene mutations at the 10p13 locus on chromosome 10, and mutations in unknown genes that result in dysembryogenesis of structures developing from the pharyngeal pouch at 8 weeks of gestation. Most cases are sporadic; boys and girls are affected with equal frequency. It is inherited in an autosomal dominant manner. DiGeorge Syndrome can be:

- ✓ Partial: Some T-cell function is present

- ✓ Complete: lack of T-cell function

Infants with DiGeorge syndrome have low ear position, median cleft face, mild mandibular underdevelopment, hypertelorism, short philtrum, developmental delay, and congenital heart disease (eg, coarctation of the aortic arch, truncus arteriosus, tetralogy of Fallot, atrial or interventricular septum). Children have hypo- or aplasia of the thymus and parathyroid glands, leading to T-cell failure and hypoparathyroidism. Recurrent infections begin immediately after birth, but the degree of immunodeficiency varies greatly, and T-lymphocyte function may improve spontaneously. Hypocalcemic seizures occur within 24 to 48 hours after birth. Carrying out an absolute lymphocyte count, followed by a B- and T-cell count and an assessment of lymphocyte subpopulations if leukopenia is detected; perform a blood test for T-cell counts and parathyroid function. Measurement of Ig levels and vaccine titers. If complete DiGeorge syndrome is suspected, a T-receptor excision ring (TREC) study should be performed. A lateral chest x-ray can help visualize thymic shadows. Fluorescence in situ hybridization (FISH) detects a deletion of the 22q11 locus on chromosome 22; standard chromosomal testing may be done to look for other disorders. Echocardiography should be done if complete DiGeorge syndrome is suspected. If patients have cyanosis, cardiac catheterization may be necessary. Because the disease is sporadic in most cases, family screening is not necessary.

Systemic (systemic) lupus erythematosus is a serious, chronic disease that affects blood vessels, skin, and connective tissue of all internal organs. This disease often occurs in young people (especially women and girls), people who are prone to the disease. In systemic lupus erythematosus, damage to internal organs plays a key role. A slow-acting virus belonging to the RNA group is the main cause of the disease. Hereditary predisposition, suffering from rheumatism and some allergic diseases, the presence of anti-measles antibodies in the blood of patients, the influence of the external environment, physical therapy procedures, sunlight, taking medications are special factors in the development of the disease. In the presence of a predisposition to this disease under the influence of the virus (sometimes under the influence of anti-viral antibodies), the management of the immunological response changes: the reduction of T-lymphocytes and the increase of the activity of B-lymphocytes - the activity of humoral immunity increases. Uncontrollable antibodies against various tissues and body proteins appear in the patient's body. In the first stage, immune complexes are formed and they settle in organs and tissues (especially in small blood vessels), these compounds injure organs and cause immune inflammation. As a result of inflammation and damage to the connective tissue, new antigens appear, in response to which antibodies are observed again, thus the disease becomes chronic. In systemic lupus erythematosus, all organs and tissues are affected. Inflammation is observed in veins and capillaries. Fibrinoid necrosis develops in the connective tissue, which is rich in fibrinoid fragmented DNA. Along with changes in blood vessels, lymphoid and plasmatic tissues are observed in the internal organs. Morphological changes in some organs are characteristic of systemic lupus erythematosus, especially peripheral sclerosis in the central artery of the spleen, wire-like nodules caused by thickening of blood vessels in the renal glomeruli, with inflammation of the heart valves and endocardium. expressed volchanka passes in the form of endocarditis.

The symptoms of the disease are different and gradually worsen. It is mainly represented by 3 "big" signs: dermatitis (inflammation of the skin), polyarthritis (inflammation of the joints), polyserositis (inflammation of various organs in the serous layer, accumulation of fluid). Systemic lupus erythematosus is a multi-symptom disease, distinguished from other diseases by its rapid development, secondary infection, and negative consequences. In the clinical picture of the disease, most of the signs dominate. The disease often begins with damage to the joints. It is called joint damage (lupus arthritis). Acute and chronic arthritis and polyarthralgias are observed in almost all patients with systemic lupus erythematosus. Fingers, palms, knees, and ankle joints are often

injured. Acute inflammation is characterized by swelling of the joints, redness of the skin, severe pain, rash, stiffness of the joints, and limited movement. There is no correlation between pain and swelling in the affected joints. Knee, ankle, wrist, paw joints are more affected. In acute lupus arthritis, permanent joint deficiency is not observed. In chronic lupus arthritis, 10-15 years after the onset of the disease, the appearance of the joints changes, ankylosis develops. Inflammation of joints is symmetrical, joint changes are noticeable or not, it is distinguished by the presence of inflammation in muscles and bones. If the joints are examined using an X-ray, signs of osteoporosis in the epiphyses of small joints, and sometimes thinning of the subchondral plate of the joint are revealed. Symptoms of acute and moderately acute synovitis can be detected by biopsy of the synovial membrane. Symptoms of the disease are found in the skin coverings as well as in the joints. Red "butterfly-like" rashes appear on the nose and around the sides. Changes in the skin have different appearances, with signs of inflammation of different severity. This inflammation increases after exposure to sunlight, cold air, and wind. The "butterfly copy" redness on the patient's face intensifies and becomes more obvious as it approaches the center, skin changes are also observed on the open part of the neck, chest, arms and legs that are not covered by clothing (flange erythema similar to fish scales). Changes in the skin are also found in the oral cavity, enanthema of the hard palate, stomatitis, chapped lips, rashes. Patients quickly lose weight, lose hair, develop baldness, brittleness of hair fibers. Nails become brittle and brittle, trophic changes and ulcers appear in the skin and mucous membranes.

Inflammation of the serous mucous membranes (polyserositis) is observed in the majority of patients with systemic lupus erythematosus. Damage to the pleura and pericardium is more common, and damage to the mucous membrane in the abdominal cavity is less common. Inflammation of the mucous membranes can be dry and with accumulation of serous, serous-fibrinous, fibrinous fluid. During echocardiography of patients, it is determined that there is serous or serous-fibrin fluid in the outer membrane of the heart. Systemic lupus erythematosus is characterized by the accumulation of fluid between the mucous membranes of various organs. Due to the short duration of serositis, when examining patients, pleuropericardial scars, focal thickening of the pleura are diagnosed. Damage to the cardiovascular system is a very characteristic feature of systemic lupus erythematosus, and in lupus carditis, the myocardium and endocardium are damaged. Patients may develop glomerulonephritis, increase blood pressure, angiopathies, damage to cerebral blood vessels. After 2-4 years of systemic lupus erythematosus, the lungs are damaged - lupus pneumonitis. Patients have symptoms such as shortness of breath, dry or sputum (sometimes mixed with blood) cough, wheezing, chest pain, moist wheezing with small bubbles in the lower lobes of the lungs, and crepitation. is represented by When examined by X-ray light, changes in the shape and increase of the lung image, high location of the diaphragm, flanged atelectasis can be seen. Sometimes lung damage leads to pneumosclerosis, pulmonary and pulmonary heart failure.

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