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CLINICAL CASE OF PATIENT WITH FAMILIAL MEDITERRANEAN FEVER

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Introduction. Familial Mediterranean fever (FMF) is a genetic autoinflammatory disorder that maker recurrent fevers and painful inflammation of the abdomen, lungs and joints. Also known as recurrent polyserositis. It is characterized recurrent episodes of peritonitis, pleuritis, and arthritis, usually with accompanying fever.

The common polymorphisms in the MEFV (Mediterranean fever) gene acting the role of pyrin in disease pathogenesis. Mutations in the *MEFV* gene are caused the disease. Gene is localized on 16p13.3 and more than 70 mutations have been described in pathogenesis. In the various ethnic groups the date of literature is described differences in the gene mutations of FMF.

Aim. To improve the diagnosis of primary immunodeficiencies in children with atypical symptoms.

Material and methods. In this article the clinical case is performed the familial Mediterranean fever in child with pulmonary manifestations.

General and medical history. A 10 years old girl is referred to a pulmonologist for evaluation of her changes in respiratory system, such as repeated pneumonia, multiple bronchiectasis, recurrent episodes of obstructive bronchitis. Heredity is negative in family history. The onset of pulmonology disorder was between the ages 2-3.

History of present illness also is included: chronic granular pharyngitis; chronic kidney disease, chronic pyelonephritis, oral mucosa candidiasis.

The patient is evaluated by the immunologist, who is primarily concerned about immunodeficiency

Standart immunological blood test is unrevealing of variations, which allowing to diagnose immunodeficiency.

In immunological blood test is identified increased levels of complement activity, CD4+-lymphocytes, T-cytotoxic lymphocytes (CD3 + CD4 + HLA-DR +), B- lymphocytes (CD19+), decreased levels of NK-cells (CD3-CD16/56+) and T-cytotoxic lymphocytes (CD3 + CD8 + HLA-DR +). Other parameters of phagocytosis and immunoglobulins A,M,G were in normal ranges.

Gene panel identifies one pathogenic variant in MEFV (c.2230G>T (p.Ala744Ser)), which is associated with autosomal recessive and dominant familial Mediterranean fever

According to genetic testing this individual is a carrier for autosomal recessive familial Mediterranean fever.

This result is insufficient to cause autosomal recessive FMF; however, carrier status does impact risk. FMF is typically autosomal recessive, and the majority of individuals with heterozygous pathogenic variants in MEFV are asymptomatic individuals, however some heterozygotes can express with mild to classic type of disease.

Conclusions. Genetic testing for identifying of pathogenic variants in MEFV is very important.

Clinical case of patient with familial Mediterranean fever/ Ezzahidi Manal, Outti Hajar, Al Saleh Ahmad, Lupaltsova O.S.// Актуальні проблеми клінічної та фундаментальної медицини ISIC,2021 р.: тези допов. - Харків, 2021. – 254 с. - С.171-172.