E131K) in exon 3 of WAS gene. The third case was a 16 years old boy who presented with thrombocytopenia and recurrent sinopulmonary infections. Immunological investigations indicated a normal immunoglobulin profile, low level of CD4(+) T cells (29.8%) and elevated CD8(+) T cells (36.6%). TREC and KREC copy counts revealed low TREC levels while KREC level was within the normal range. Molecular analysis of the WAS gene revealed two mutations – c.57 G>T (p. Q19H) in the first exon, and c.136 C>A (p. L46M) in the second exon. The presumed impact on the patient phenotype was investigated on The Ensembl Variant Effect Predictor and as a result, c.57 G>T (p. Q19H) mutation has a severe phenotypic effect, while the impact of c.136 C>A (p. L46M) mutation is moderate.

Prospects for further research. In order to improve the diagnosis of primary immunodeficiency disorders and to achieve an efficient differential diagnosis, we intend for the future to broaden the spectrum of molecular-genetic diagnosis of immunodeficiency diseases in the Republic of Moldova.

References:

Key words: primary immunodeficiency, Wiskott–Aldrich syndrome, thrombocytopenia.

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ABOUT THE BIOMARKERS OF THE TRANSITION OF HUMAN INTERVERTEBRAL DISK FROM THE NORM TO PATHOLOGY

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It is known that tissue of intervertebral disc (ID) is characterized by intensive glycolysis, the marker of which may be components of the lactate dehydrogenase (LDH) system. As known, ID receives nutrients and O2, and also carries out the outflow of metabolites by diffusion with use of vertebral bodies. Dystrophic-degenerative changes in ID, which can occur during stress, are the basis of osteochondrosis (OCh). The results of the experiment on white rats or different ages showed that during the response to stress, the components of the LDH system (total activity and isoenzyme spectrum of LDH, lactate, pyruvate content and their ratio) in the spinal tissues are characterized by phase changes. They consist in the switching of glycolytic processes in ID to the activation of aerobic metabolism, which may correspond to the depletion stage (according to G. Selye's theory of stress). If it is necessary to carry out an operation for a patient with osteochondrosis of the spine, we obviously deal with the depletion stage. It can not always be characterized by instrumental methods of examination. Therefore, it is relevant to search for biochemical markers of the transition of human ID in a pathological condition. The purpose of this work was to study the components of the LDH system in human ID tissues obtained during spinal cord surgery in OCh, as well as ID cadaveric material, to characterize the intensity of glycolysis in this tissue during its transition from norm to pathology.
**Materials and Methods.** Studies were carried out in homogenates of ID tissues in patients with OCh of 45-55 years after surgery and cadaveric material of healthy men of the same age after a car accident, which was used as a control. The total LDH activity was determined by a spectrophotometric method based on the Warburg optical test, the isozyme spectrum of LDH by the Davis polyacrylamide gel electrophoresis method, the determination of lactate and pyruvate by the enzymatic method Rollinghoff. Statistical data processing was performed using Student’s T-test. The results of the study showed that the ID tissue of patients with OCh is characterized by a decrease of almost 3 times as compared with the control by the lactate/pyruvate ratio due to a decrease in the lactate content and pyruvate accumulation. At the same time, the total LDH activity in ID patients with OCh was reduced by 50-60% compared with control values, and the isoenzyme spectrum of LDH was shifted towards an increase in the LDH2 isoenzyme. These results indicate that during the development of OCh, the metabolism of ID switches from anaerobic, characteristic of this tissue to normal, to aerobic, which has its own anatomical explanation. When pathology in the ID, microvessels sprout and it switches from the diffuse type of nutrition to the production of nutrients and O2 and the outflow of metabolites through the vessels.

The prospect of the results of the work can be the use of glycolysis markers in combination with the determination of the components of the extracellular matrix of the connective tissue ID to develop quantitative biochemical criteria for its pathology. The source of optimism for the prospects is the experimental data that revealed a high degree of correlation of changes in the content of lactate, pyruvate, the value of the lactate / pyruvate coefficient in ID and the blood of animals in the older age group under various stress effects.

**References:**


**Key words:** intervertebral disc, markers of glycolysis, degenerative changes, stress

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**THE ROLE OF GASTROINTESTINAL DAMAGE DIAGNOSTIC BIOMARKERS TO IMPROVE POSITIVE COMPLIANCE FOR ANTIVIRAL THERAPY OF HIV-POSITIVE PATIENTS**

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Ukraine is currently one of the first countries in the European region in terms of the number of human immunodeficiency virus (HIV)-positive people. It is important to carefully follow antiretroviral therapy (ART) regimens for the management of such patients [1]. At the same time, the incidence of abdominal pain in the HIV- patient population ranges from 12 to 45 percent [2]. The often causes of such pain in HIV-infected patients are peptic gastroduodenal ulcers (PGDU) caused by Helicobacter pylori infection (HP) and the use of non-steroidal anti-inflammatory drugs (NSAIDs) [3].