

Conclusions. In this research, we found that 42% of the patients with obesity and intellectual deficiency were carriers of pathogenic genetic abnormalities that can explain their symptoms. Although some of the patients presented classical variants described in literature, some of our findings are variants that were not previously described or were described in very few cases.

Key words: obesity, Intellectual, developmental, copy number variants

275. PRENATAL DIAGNOSIS OF CONGENITAL MALFORMATIONS OF THE BRAIN IN PREGNANCIES WITH GENETIC RISK

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Introduction. The medical-genetic counseling is one of the most widespread and effective methods of prenatal diagnosis (PD) and prophylaxis of congenital and hereditary pathologies.

Aim of the study. To highlight the role of medical-genetic counseling and prenatal diagnosis in pregnancies with risk for malformations of the brain (MB) at early stages of intrauterine development to reduce the incidence of congenital MB in newborn.

Materials and methods. The medical-genetic counseling of the 657 pregnant women during 2015-2017 years, which were divided into two groups: a) I group - 239 women with medium and high genetic risk; b) the II group - 418 women with low genetic risk.

Results. All pregnant women in the study performed noninvasive PD: ultrasound and biochemical screening. In 49 cases the values of serum alpha-fetoprotein were elevated. Examination of pregnant women on informative terms by non-invasive prenatal diagnosis (fetal ultrasonography) allowed the diagnosis of MB to fetuses in 33 cases. Cerebral fetal malformations diagnosed prenatally through the ultrasound examination were: spina bifida - 6 cases, anencephaly - 5 cases, holoprocencephaly - 5 cases, corpus calosum agenesis - 7 cases, hydrocephaly - 4 cases, Dandy-Walker malformation - 3 cases, schizencephaly - 1 case, lissencephaly - 1 case. The medical-genetic counseling were provided to couples. The final decision to interrupt the pregnancy was made by couples. A prophylaxis plan was developed in families with genetic risk.

Conclusions. PD and medical-genetic counseling help to reduce the frequency of congenital malformations in newborns also makes it possible to prevent the birth of children with CM and chromosomal abnormalities diagnosed prenatally until 21 weeks of gestation.

Key words: congenital malformations of the brain, prenatal diagnosis, medical-genetic counseling

276. MULTIPLE EXOSTOSIS - CAUSES AND POLYMORPHISM

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Introduction. Multiple exostosis is a genetic bone disease characterized by the development of osteochondroms present in the form of long-bone bony bumps. These bone-to-bone bumps have different shapes and are formed in restricted populations whose populations suffer from mutation in chromosome 8 manifested by the lack or insufficiency of the exostosin-1 protein.

Aim of the study. The purpose of this study was to present the. Clinical and genetic study of multiple exostosis, moreover the correlation between the genetic and clinical aspects of the disease.

Material and methods. The study includes the experience of surgical treatment of 11 patients, clinically confirmed, radiologically, morphologically confirmed in the conditions of the orthopedic and traumatology clinic of "N. Testemitanu".

Results. The difference in localization of the pathological outbreak, on the right or left side, is almost equal. The solitary formations in 73.03% were located on the humerus, tibia and phyllus and 26.97% in the other bones of the skeleton: the humerus - 1, the radius - 2 ulna - 1, the femur - 3, the tibia - - 2, calcaneus - 1. Pathological focal areas of the clavicle, scapula, humerus, radius, ulna, femur, tibia, fibula, cuneiform bone, calcaneum were removed in patients with multiple pathology as indicated. The particularities of this disease usually allow sparing surgery - marginal resection in the affected bone region (93%), without the need for osteoplasty, and only 7% performed other types of resection. In all cases, the resection piece was studied patomorphologically in a specialized laboratory.

Conclusions. Given that the prevalence of the disease in Moldova according to Ministry of is estimated at 1:35 000 individuals it has been difficult for me to analyze the patients because the vast majority of them are operated and in short type are dispensed. At the same time, we noticed that a patient had relapses, so the number of surgeries a patient needs during his life varies from one person to another. Treatment of the disease is by removing very bulky exostoses, which causes pain, joint limitations, nerve or vascular compressions or massive bone deformities. A careful follow-up of the affected person allows to determine the optimal moment of intervention and prevent complications such as joint dislocations. With some issues related to unsightly appearance or reduced functionality of some skeletal segments, this disease is compatible with normal life.

Key words: exostosis, polymorphism, humerus

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277. SOME ASPECTS OF ADRENAL STRUCTURE AND FUNCTIONING IN THE FETUS

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Introduction. In adults, the adrenal gland consists of two parts: the outer one - the adrenal cortex and one in the inside - the medullary adrenal. The epithelial cortices of the adrenal cortex are arranged in three areas: the glomerular area, which specializes in the production of mineralocorticoids (the main representative being aldosterone), the fasciculating area producing glucocorticoids (cortisol, cortisone) and crosslinked area possessing the ability to deliver sex hormones androgens, estrogens).

Aim of the study. Description of physiological and histological changes occurring in the adrenal glands of the fetus.

Conclusions. The adrenal glands have an important role in the prenatal period, contributing to the development of the fetus as well as throughout life. From a histological point of view, the adrenal glands at the fetus have a different structure from the adult, which is represented mostly by permanent and temporary fetal cortical and a disorganization of the medullary substance.

Key words: adrenal gland, cortex, medullar.