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 RISC

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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, holoprozencephaly - 5 cases, corpus calosum agenesia - 7 cases, hydrocephaly - 4 cases, Dandy-Walker malformation - 3 cases, schizencephaly - 1 case, lisencephaly - 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 osteocondrograms present in the form of long-bone bosel These bone-to-bone bumps have different shapes and are formed in restricted populations whose populations suffer from mutation in comosome 8 manifested by the lack or insufficiency of the exostosin-1 protein.