The aim of this study was to estimate the prevalence of fetal cardiac anomalies in the first trimester of pregnancy in pregnant women with high degree of genetic risk.

Methods: We analyzed data regarding ultrasound examination, the nuchal translucency, visualization of the four-chamber view, the outflow tracts, double test measured in first trimester of pregnancy, in 128 pregnant women who have been investigated for medico-genetic counseling in 2009-2010.

Results: In 44 (34,4%) pregnant women (average age 26,1 ± 5,3 years) was estimated medium degree of genetic risk, in 30 (23,4%) - high risk and in 54 (42,2%) - low risk. Prenatal diagnosis has contributed to the identification of severe fetal pathologies in 16 (12,5%) pregnant women. The most common cardiac defects included 6 atrial and 2 ventricular septum defects (37,5% and 12,5% respectively), anomalies of the aortic arch or its major branches 5 (31,3%), D-transposition of the great arteries in 3 (18,5%) cases. Amniocentesis with the study of fetal karyotype allowed the identification of numerical and structural chromosomal abnormalities in 18 patients (14,0%), in 2 of them were detected structural chromosomal abnormalities with 22q chromosome.

Conclusions: Investigation on methods of primary prevention prenatal diagnosis (fetal ultrasound, karyotyping) is essential to reduce the frequency of chromosomal abnormalities and congenital malformations.

Key words: velocardiofacial syndrome, chromosome, prenatal diagnosis.

CADASIL

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Introduction: Cerebral autosomal dominant arteriopathy with subcortical infarctions and leukoencephalopathy (CADASIL) is a single gene disorder of the cerebral small blood vessels caused by mutations in NOTCH3 gene. The disease has been recently described and the exact prevalence is unknown currently, but the number of the reported cases is increasing as the clinical picture is better known due to active research work in this field. The main clinical manifestations are recurrent stroke, migraine, psychiatric symptoms, and progressive cognitive impairment. The clinical course is highly variable, that's why the disorder is often misdiagnosed. The pathological hallmark of the disease is the presence of granular osmiophilic material in the walls of the affected vessels, which can be detected in skin biopsy. The diagnosis is important as the clinical course and the prognosis differ between patients with CADASIL and those with other common cerebral small vessel diseases. Moreover, the usual therapy for ischemic stroke, which includes thrombolytics, antihypertensive agents and statins, has been not validated for CADASIL patients.

Conclusion: In the Republic of Moldova was described one family affected by this disease. I will present a family tree which includes three generations, persons that suffered of this disease and age at which the main manifestations appeared. As very little is known about the disease, I think my presentation will clarify how the disease occurs and what can be done and, not less important, will call attention on this issue.

Key words: Record Card, statins, antihypertensive agents.