

Prenatal diagnostics is headed in several directions - towards visualization of fetuses and biochemical, cytogenetic and molecular genetic diagnostics in laboratories.

Whereas visualization of fetuses does not a priori represent any direct risk for pregnancy and does not increase the number of potential pregnancy complications, this is not always the case with the laboratory testing. There are known risks connected with invasive methods of prenatal diagnostics.

The number of potential unintentional pregnancy complications and losses as well as the technical and economic aspects of invasive prenatal diagnostics lead to attempts of identifying ways of detecting any potentially affected individuals by screening methods, thus minimizing the undesirable impact of invasive diagnostics on the pregnant population. The more precise the selective criteria, the lesser the number of pregnant women exposed to invasive exams.

Another way of decreasing the number of unintentional complications in relation to invasive diagnostics is to simplify and improve the fetal samples harvesting methods during pregnancy.

The work primarily focused on two areas: Determination of the relation between fraction shortening of the left and right ventricles and a fetal chromosomal complement, and verification of reliability of a new method of amniotic fluid and chorion villus sampling using new vacuum tubes.

We have confirmed that it is possible to routinely measure functional parameters of the fetal heart as early as towards the end of the first trimester of pregnancy and that the measuring results may be used to distinguish between the aneuploid fetuses and the fetuses with normal karyotype. We have identified differences in fraction shortening values in euploid and aneuploid fetuses. Our measuring further suggests that potential etiology of tricuspid regurgitation in the first trimester of pregnancy is an enlarged right ventricle.

In the second part of the work, we have proved that the method of harvesting samples of amniotic fluid and performing chorion villus sampling, using vacuum tubes developed by us, is reliable and safe.