



P164. TRISOMY OF X CHROMOSOME AND DIAGNOSIS

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Until about a decade ago, it was not possible to imagine that non-invasive methods could so precisely detect very reliable genetic changes on chromosomes, even the finest (microdelecia) ones, while today these methods occupy a significant place in prenatal screening. The contribution to avoiding the risk of using invasive prenatal diagnostic procedures is well known to all of us, especially considering that it is quite common (at least in Serbia) that women refuse to do this type of diagnosis just because of the existing risk.

We wanted to show the sensitivity and specificity of non-invasive prenatal testing in the case of a very rare numerical aberration of the sex-determining X chromosome, in case it is indicated due to the age of the pregnant woman.

Pregnant woman aged 36 years, second pregnancy. The first one was normal, the child was healthy, no prenatal diagnosis was performed because there was no indication. Mother and father with no history of genetic anomalies. Non-invasive prenatal diagnosis done due to the risk that exists in the invasive diagnostics that is per protocol for her age. The obtained result shows the trisomy of the sex-determining X chromosome in the analyzed material. Considering that in Serbia only the invasive diagnostic is valid for granting pregnancy termination, pregnant woman is referred to the amniocentesis that confirmed the diagnosis. A pregnancy terminates in 19 weeks (the amniocentesis result is waited for 3 weeks). Fetal autopsy confirmed trisomy of X chromosome.

We know that the trisomy of the X chromosome is rare in relation to all other numerical aberrations, both in sex-determining and autosomal chromosomes. Based on this case, we can see how high the sensitivity and specificity of the non-invasive prenatal test is and how significant a change would be if it was accepted to approve a termination of a pregnancy. Pregnancy could be terminated by week 14, taking into account the time when the test is done and how long it takes to reach the results. This would avoid the complications that exist in instillation when the results of amniocentesis are reached, as well as the time of hospitalization of the pregnant woman in the event of a pregnancy termination at week 14.

We can say that the priceless advantages of non-invasive prenatal test are that it is non-invasive, that it is done in the first trimester of pregnancy, that the results arrive in 7-10 days and that, in case of poor findings, it is possible to perform termination of pregnancy much earlier than with amniocentesis. It should never be neglected that its specificity and sensitivity is over 99%. Hopefully, in the near future,

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the price will be more affordable and most countries will be able to make it a regular testing when there are indications.