Patient Information Series – What you should know, what you should ask.

What is tricuspid regurgitation?

The tricuspid valve is one of the two valves between the upper (atria) and lower (ventricles) chambers of the heart. It is located on the right side (between the right atrium and the right chamber) and it is call tricuspid because it contains three leaflets that close before the heart contracts. When the closure is not tight, some blood leaks from the chamber backwards towards the atrium. If the leakage is severe, we call it "regurgitation". This feature is seen more often in babies with a chromosomal anomaly, especially Down syndrome (extra chromosome 21) or with a heart defect and therefore when seen, we call it a marker for these abnormalities, and it increases the risk of them being present in your baby.

However, TR can also be seen in normal babies, and it is considered a finding without a clear explanation. In screening tests for Down Syndrome (such as the combined test), the finding of tricuspid regurgitation (TR) increases the chance that your baby has a problem with the chromosomes above the initial risk calculated by the combined test. The doctor will discuss with you what the risk given to you means and help you interpret the test result. For instance, a risk of 1:100 is as "high" and means that if 100 women have been given the same risk, only one of them will have a baby with a problem and 99 will not.

Ultimately, interpretation of risk is highly individual and only you can decide which risk is acceptable for your and whether you wish to follow up a positive screening test with a confirmatory, diagnostic test such as taking a small sample from the placenta or some amniotic fluid from around the baby (chorion villous sampling/amniocentesis) to analyze the chromosomes.

Should I have the non-invasive test (NIPT) done?

Your doctor will advise you if you qualify for the NIPT or rather should consider having an invasive test

How are chromosomes relevant to tricuspid regurgitation?

Chromosomes are where most of our genetic information is kept. We usually have 46 of them matched in pairs: 23 come from one parent and the other 23 come from the other parent. For example, people with Down syndrome have an extra chromosome number 21. Babies with a chromosomal anomaly, usually an extra chromosome 21 or 18 or 13, often show a thickened NT and/or sometimes show tricuspid regurgitation. This can also happen when the baby has a heart anomaly, even if the chromosomes are normal



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Should I have more tests done?

If the result of the diagnostic test is normal, the doctor will probably suggest you doing an **advanced fetal scan** to analyse the all the fetal structures, but in particular, the heart to check if this is normal.

What are the things to watch for during the pregnancy?

If the tests are all negative (chromosomes and scan of the heart of the baby) it means that no problems have been found with your baby and your pregnancy can be considered normal, so you do not need any further special care in pregnancy. The tricuspid regurgitation will also disappear at a later stage in pregnancy and the explanation for the fact that it was visible early in pregnancy is not clear.

Will it happen again?

Usually, TR will not happen again in the next pregnancy, but if the baby had a chromosomal anomaly the risk may be slightly increased in your next pregnancy.

What other questions should I ask?

- What is the chance of my baby having a chromosomal anomaly if there was TR?
- Can the doctor do a combined test and give me an exact risk of chromosomal anomalies in my baby?
- Which invasive test should I opt for (chorion villous sampling or amniocentesis)?
- Which risks are associated with these invasive tests?
- Which genetic investigations will be performed?
- Will these tests diagnose small genetic problems?
- Will the doctor organize extra specialized scans in the pregnancy to check if the TR was associated with a heart problem?
- If both heart and chromosomes are normal, will my baby be checked later in the pregnancy to see if the TR has disappeared?

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