Absent nasal bone in the baby at 11-14 weeks

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

What is an absent nasal bone?

The **bone present in the nose** of your baby should already be developed **between 11 and 14 weeks'** gestation and be visible at the ultrasound scan performed at this time in pregnancy. The nasal bone appears on a profile view of the fetal face as a thin white line. It has been observed that when the nasal bone is not visible there is an increased chance that the baby may have a chromosomal anomaly, such as Down syndrome (extra chromosome 21) or other chromosomal anomalies.

The presence/absence of the nasal bone may also be used to improve the risk calculation of your baby having a chromosomal problem and improve the accuracy of a standard screening test, such as the **combined test.** This test combines the measurement of the nuchal translucency of the baby with pregnancy hormones measured in your blood and calculates the risk of a chromosomal anomaly in your baby.

There are variations among countries, but usually a risk equal or higher than 1:100 is considered high. The doctor will discuss with you what the risk means and help you interpret the test result. Ultimately, interpretation of risk is highly individual and only you can decide which risk is acceptable for you 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 such as amniocentesis or CVS.

How are chromosomes relevant to an absent nasal bone?

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 a thin or absent nasal bone. This can also happen when the baby has a very small genetic defect, at the gene level. That is why your doctor may suggest doing an even deeper examination of the genetic material of your baby.

In women of Afro-Caribbean origin the development of the nasal bone in the baby is slower and therefore there is a higher chance that it may not yet be visible at 11-14 weeks.

Absent nasal bone in the baby at 11-14 weeks

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

Should I have more tests done?

If the result of the diagnostic test is normal, the doctor will probably suggest you do an **advanced fetal scan** to analyse all the structures of the fetus for structural anomalies.

What are the things to watch for during the pregnancy?

If the tests are all negative 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 nasal bone will become visible at a later stage in pregnancy and the explanation for the fact that it was not visible early in pregnancy is that probably its development was slower than in other pregnancies, without any special reason.

Will it happen again?

Usually, an absent nasal bone 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 its nasal bone is not visible?
- Can I have a combined test done and get my baby's calculated risk of having a chromosomal anomaly?
- Do I qualify for a non-invasive blood test?
- If the risk is high which invasive test should I choose (chorion villous sampling or amniocentesis)?
- What is the risk associated with these invasive tests?
- Which genetic investigations will be performed?
- Will these tests diagnose other small genetic problems?
- Will the doctor organize extra specialized scans in the pregnancy to check if the nasal bone has become visible later in pregnancy and if there are other anomalies?

Last updated April 2023