Absent Nasal Bone

What is meant by absent nasal bone?

It refers to a finding on the ultrasound where the baby's nose bone appears absent or smaller than normal.

What does absent nasal bone signify?

It is important to know that even in normal babies, the nasal bone is absent in about 1-3% cases. However, studies show that in about 40 - 60% babies with chromosomal abnormalities the nasal bone may be absent or may appear later than normal. Hence it warrants evaluation of the baby's chromosomes.

When to assess on ultrasound?

It is best to assess at around 11th to 14th weeks of pregnancy (1st trimester). If nasal bone is absent at this period then it should be reassessed at around 16th to 17th weeks of gestation.

Does this mean that the baby would be born without a nose or a malformed nose and disfigured face?

No, the baby will not have a disfigured face. It will have a nose but may be smaller than normal.

How is the risk for chromosomal abnormalities in the baby assessed in the presence of absent nasal bone?

Characteristics like your body mass index, whether there is a history of previous babies with chromosomal abnormalities etc. will be entered into a computer software.

Integrating the above parameters and other ultrasound scan findings, the computer software will generate a risk number. Any risk number of more than 1: 250 is considered 'high risk'. For example: 1: 75 - which means that

if there are 75 mothers having the same parameters, then 1 in 75 such mothers have a risk of having a baby with chromosomal abnormality.

What other tests can be done to further assess the risk of having a chromosomally abnormal baby?

You have the following options to further assess the risk of having a chromosomally abnormal baby

- Invasive testing (Chorionic villus sampling) which is the only test to know 100% whether your baby has chromosomal abnormality or not. However, it carries a procedure related risk of miscarriage of 1:300

- Non invasive maternal cell free fetal DNA -to assesses the fetal DNA in the maternal blood. It has a detection rate of 99%. If the results are "negative" then very unlikely that the baby has a defect in chromosomes 21, 18 or 13. However, if it is reported "positive" then this will need to be further confirmed by invasive testing

- First trimester Quadruple marker test - which is a blood test that measures four hormones in the mother's blood. It increases the detection rate for chromosomal abnormalities to about 90%

- Sequential screening - Combining the first trimester Combined test (NT + first trimester quadruple marker test) with second trimester quadruple test done at 15 - 16 weeks with Genetic sonogram (anomaly scan) done at 18 - 19 weeks. This has a detection rate of about 95% but does not confirm whether your baby has any chromosomal abnormality or not

What is the further course in the pregnancy?

If the risk for having a chromosomal abnormality in your baby is low or if on invasive testing it is found that the chromosomes of the baby are normal,then you will be called for follow up scan at 18-19 weeks to assess if the baby has any structural problems .