LEARN MORE ABOUT

ABSENT OR SHORTENED NASAL BONE

WHAT IS AN ABSENT OR SHORTENED NASAL BONE?

During development, the left and right nasal bones meet in the middle and become calcified to form a single nasal bone. This bone typically looks like a thin white line on ultrasound. The length of this nasal bone varies and is dependent on many factors, such as the gestational age of the pregnancy and the ancestry of the biological parents. Sometimes, the nasal bone is shorter than expected, or the bone may not be seen at all. A short or absent nasal bone is not a birth defect. However, it is an ultrasound finding that occurs more commonly in children with a chromosomal difference, such as Down syndrome.

HOW COMMON IS THIS FINDING?

1-3% of all pregnancies have this ultrasound finding

COULD AN ABSENT OR SHORTENED NASAL BONE MEAN THAT THE BABY HAS OTHER HEALTH CONCERNS OR BE ASSOCIATED WITH CERTAIN CONDITIONS?

In many cases, a shortened or absent nasal bone can be normal variation. It is estimated that approximately 1-3% of babies with the typical 46 total chromosomes will have an absent nasal bone on prenatal ultrasound. However, this finding is seen more commonly in pregnancies with Down syndrome. At 11-14 weeks gestation, the nasal bone is absent in about 66% of babies with Down syndrome and at 15-24 weeks gestation, the nasal bone is absent in about 40-50% of babies with Down syndrome. Down syndrome is caused by an extra chromosome 21 and is associated with intellectual disabilities and other health concerns. An absent nasal bone has also been reported in pregnancies with other chromosomal differences that have additional medical issues, such as monosomy X (Turner syndrome), trisomy 18 (Edward's syndrome), and trisomy 13 (Patau syndrome).

DO I NEED TO CONSIDER ADDITIONAL TESTING?

Based on your ultrasound and other testing results, your genetic counselor and/or prenatal care provider may be able to tell you more about the specific risks for your pregnancy given this finding. You may be offered genetic testing, including non-invasive prenatal testing (NIPT) or a diagnostic procedure (chorionic villus sampling [CVS] or amniocentesis), to better assess if there could be a chromosomal cause for the absent or shortened nasal bone. Prenatal genetic testing is always optional; some families may find certain genetic tests helpful for delivery preparation or for decision- making, while other families may not want additional information or may not be comfortable with testing. It is important to know that there are no tests available to detect all health conditions.

Ultrasound

Non-Invasive Prenatal Testing (NIPT)

Diagnostic testing

CAN THIS CAUSE PROBLEMS FOR THE PREGNANCY?

An absent or shortened nasal bone does not cause any health concerns to the baby itself and is not considered a birth defect. The baby will still have a nose and be able to breathe. However, a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy, such as a level II ultrasound at 20 weeks that screens for other physical features associated with Down syndrome.

WHERE CAN I GET MORE INFORMATION?

If you have additional questions or concerns about this ultrasound finding, please don't hesitate to contact your genetic counselor and/or prenatal care provider. Your healthcare team is here to help you during pregnancy.

WANT MORE INFORMATION ABOUT FINDING A GENETIC COUNSELOR? VISIT WWW.ABOUTGENETICCOUNSELORS.ORG