

What is Anencephaly?

Anencephaly is a severe birth defect in which a fetus develops without major parts of the brain, such as the cerebrum and cerebellum, as well as a significant portion of the skull. It is a type of neural tube defect (NTD) that occurs during the first month of pregnancy, often before a woman knows she is pregnant. Unfortunately, anencephaly is always fatal: the baby cannot survive long after birth. Anencephaly differs from Acrania, a rare malformation where the skull is partially or completely absent, but there is a normal or near-normal brain, typically covered only by a thin layer of membrane.

What Causes Anencephaly?

The exact cause of anencephaly is unknown. However, it is believed to result from a combination of genetic and environmental factors. These may include nutritional deficiencies, particularly a lack of folic acid, genetic mutations, and certain environmental exposures. It is highly recommended that women planning to become pregnant take a daily supplement of folic acid to help prevent neural tube defects.

Should I Have More Tests Done?

Anencephaly is diagnosed by prenatal ultrasound, which can show missing parts of the brain and bones of the skull. Anencephaly can be diagnosed at the first scan that is performed during pregnancy. You can consult with your caregiver and/or a maternal-fetal medicine specialist to decide whether terminating the pregnancy is the right choice for you and your family. Genetic counseling and testing could be recommended. This may include tests like amniocentesis or chorionic villus sampling (CVS) to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing.

What Are the Things to Watch for During My Pregnancy?

During pregnancy, regular check-ups are crucial. If anencephaly is diagnosed, your doctor might monitor your pregnancy more closely if you decide to continue. You should notify your healthcare provider immediately if you notice any unusual symptoms or changes in your health.

Anencephaly

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

What Does It Mean for My Baby After It is Born?

Infants born with anencephaly are usually blind, deaf, unconscious, and unable to feel pain. While some may be born alive, the lack of a major portion of the brain and skull leads to the baby's death within hours or days after birth.

Will It Happen Again?

The risk of having another pregnancy affected by anencephaly or another neural tube defect is increased. You may be referred to genetic counseling to discuss your risks and testing options for future pregnancies. Taking folic acid supplements before and during early pregnancy can significantly reduce this risk.

What other questions should I ask?

- What support services are available for families experiencing this diagnosis?
- Are there implications for future pregnancies, and how can I manage them?
- What treatment and care options are available during and after the pregnancy?
- How can I and my family receive psychological support during this time?

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