Anencephaly

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

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. 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?

It is a type of neural tube defect (NTD) that occurs during the first month of pregnancy, when the top part of the baby's spinal cord does not close properly. In most situations, the exact cause is not known, but it is believed to result from a combination of genetic, nutritional, and environmental factors, such as lack of folic acid intake, taking folic acid antagonist drugs, maternal obesity, diabetes, and others. 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. Other anomalies may be present, such as spina bifida, cleft lip/palate, urinary tract and gastrointestinal anomalies, heart defects, and polyhydramnios. The alpha-fetoprotein (AFP) level in maternal serum could also be increased. 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.

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

Unfortunately, anencephaly is a fatal congenital birth defect. Most babies with anencephaly die in the uterus, and 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. Infants born with anencephaly are usually blind, deaf, unconscious, and unable to feel pain.

Will it happen again?

The risk of recurrence, having another pregnancy affected by anencephaly or another neural tube defect is increased, and may be up to 2%. You may be referred to genetic counseling to discuss your risks and testing options for future pregnancies. Taking folic acid supplements before and early in pregnancy can reduce this risk significantly. It's recommended that all women who can get pregnant take a daily supplement containing 0.4 to 0.8 milligrams of folic acid. A higher dose of 4mg daily may be recommended for those at higher risk.

What other questions should I ask?

- Is my baby with anencephaly able to feel pain?
- Is there a genetic basis to my baby’s anencephaly?
- What treatment and care options are available during and after the pregnancy?
- What are my risks for future pregnancies, and how can I manage them?
- When should I see a healthcare provider if I want to get pregnant again?
- What support services are available for families experiencing this diagnosis?

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