# Anencephaly

BIRTH DEFECT RESEARCH FOR CHILDREN

## What is Anencephaly?

Anencephaly is a type of neural tube defect (NTD). The neural tube is a hollow tube that folds and closes during embryonic development to form the brain and spinal cord. Anencephaly occurs when the neural tube fails to close properly which leads to the absence of major portions of the brain, the skull, and the scalp. The existing portion of the brain is often exposed. Although babies with anencephaly have extremely deformed heads, they usually have normal bodies and functioning organs. The presence of a brain stem can keep the organs operating for a period of time, but the condition is ultimately fatal.

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## How Many Children Have Anencephaly?

Anencephaly occurs in one out of 1,000 live births, affecting females more often that males. It occurs six times more frequently in Caucasian babies than in African-American babies.

## How Do You Know If Your Child Has Anencephaly?

Anencephaly is observable at birth. Three prenatal diagnostic techniques can help to determine whether a baby has anencephaly. Pregnancies in which the baby has an NTD show an elevated level of a substance called alpha-fetoprotein (AFP). An AFP screening test measures the amount of AFP in the mother's blood. Amniocentesis (a test of the fluid surrounding the baby) may also show an elevated AFP level and an increased amount of an enzyme called acetylcholinesterase. There are risks associated with this test (such as puncture to the baby and possible miscarriage) which you should discuss with your doctor. Anencephaly can also be detected by ultrasound. Any of these three prenatal diagnosis techniques should be performed between 14 and 18 weeks of pregnancy.

## What Causes Anencephaly?

Anencephaly results from a defect in the closure of the neural tube occurring between 16 and 26 days after conception. NTDs are in a category of birth defects called polygenic or multifactorial. This means that they are caused by one or more genes interacting with an environmental factor. Environmental triggers for NTDs that are being studied include vitamin and mineral deficiencies, chemical exposures, drugs, viruses, and maternal illnesses The recurrence rate for anencephaly is 4-5% and increases to 10-13% if the parents have had two previous babies with anencephaly. If one parent has an NTD, the risk of having a child with an NTD is 3-5%. In these situations, genetic counseling is recommended. Folic acid supplementation prior to and during pregnancy may reduce the risk of recurrence.

## How Can You Help A Child With Anencephaly?

There is no cure or major medical intervention for a baby with anencephaly. The treatment is usually supportive and includes keeping the baby warm, protecting the exposed brain tissue, and feeding the baby. A special bottle called a Haberman Feeder can make it easier to feed a baby who has a sucking motion but has difficulty swallowing.

## What's In The Future For A Child With Anencephaly?

Anencephaly is a fatal condition. About one-half of babies with anencephaly are born alive. Of these babies, 25% live 3-5 days, 50% live up to 10 days, and the remaining 25% live up to 15 days. In a few rare cases, babies with anencephaly have lived several months.

Fact Sheet by:

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