ANENCEPHALY- A NEURAL TUBE DEFECT IN FOLIC ACID DEFICIENT PREGNANT WOMAN

Pathak Meenakshi S.N.¹ and Awadhesh Pandey²*

¹Assistant Professor, Department of Prasuti Tantra, Patanjali Bharteeya Ayurvigyan Avem Anusandhan Sansthan, Haridwar Uttrakhand.
²Assistant Professor, Department of Shalya Tantra, Patanjali Bharteeya Ayurvigyan Avem Anusandhan Sansthan, Haridwar Uttrakhand.

ABSTRACT

Neural tube defects are birth defects that affect the tissue that grows into the spinal cord and brain. Among them Anencephaly is one of the most common neural tube defects found in folic acid deficiency during pregnancy. Anencephaly occurs when the upper part of the neural tube fails to close. Possible causes include environmental toxins and low intake of ‘folic acid’ by the mother during pregnancy. Having one infant with this condition increases the risk of having another child with neural tube defects. Folic acid is an essential supplement to be advised in pregnancy to prevent such neural tube defects. The preconceptional use of folic acid containing supplements reduces the first occurrence, as well as the recurrence, of neural tube defects. Such deficiency during the first four weeks of gestation can result in structural and developmental problems. Folate intake may need to be sustained after complete closure of the neural tube to decrease the risk of other poor pregnancy outcomes. A central feature of embryonic and fetal development is widespread cell division; folate is central because of its role in nucleic acid synthesis. Folate deficiency can occur because dietary folate intake is low or because the metabolic requirement for folate is increased by a particular genetic defect or defects. Women of populations in which adverse pregnancy outcomes are prevalent often consume diets that contain a low density of vitamins and minerals, including folate.

KEY WORDS: anencephaly, counselling, folic acid, preconceptional, pregnancy.
INTRODUCTION

The literal meaning of anencephaly is "no brain." But the term is not exactly accurate for describing the condition of anencephalic babies. (Image 1)

\[ an = \text{negative} \quad \text{enkephalos} = \text{brain} \]

Anencephaly is a common congenital disorder in which, during the 21st to 28th day after fertilization, the neural tube of the fetus does not close; therefore, the fetus is missing its prosencephalon (also called the forebrain), and large parts of its skull and scalp. The prosencephalon, which includes the diencephalon and the telencephalon, is responsible for the development and use of speech and language, motor functions, olfactory senses, memory, emotion, certain metabolic processes, and autonomic activities. The areas of the brain which are formed in an anencephalic fetus are left exposed, without any skin or bone to cover it as seen in the Image 2.

Such babies have no cerebrum or cerebellum but they do have a brain stem. The brain stem allows them to breathe and allows their hearts to beat. But the babies cannot see, hear, or feel.

Epidemiology

Anencephaly occurs in about 1 out of 10,000 births. The exact number is unknown, because many of these pregnancies result in miscarriage.
Other neural tube defects

- spina bifida (especially cervical)
- congenital heart defects
- cleft lip/palate
- diaphragmatic hernia
- spinal dysraphism
- skeletal anomalies: e.g. clubfeet
- gastrointestinal abnormalities: e.g. omphalocele
- urinary tract abnormalities: hydronephrosis most common

Causes

1) Folic acid defiency (Which can be caused by poor or inadequate diet, smoking, alcohol use, psoriasis, & birth-control pill use)\[^5\]
2) Undiagnosed diabetes
3) Hypervitaminosis A
4) High temperatures of 102 degrees or higher for more than 5 hours. (Hot tub use is discouraged when pregnant)
6) Anticonvulsant medication, especially Valporic acid (valporate)
7) Environmental/chemical exposure
8) Rare genetic cause (Genetics have long been considered likely because of the increased risk of having a 2nd child with anencephaly, but scientists still remain uncertain about the evidence to prove or disprove this theory. Some genetic disorders such as waardenburg syndrome and certain ethnic groups seem to have a higher incidence of anencephaly.)

Risk Factors

The risk factors that can increase the chance of having a baby affected by anencephaly are such as genes, behaviors, and things in the environment.\[^6\] Any woman with a family history of neural tube defects.

Low intake of folic acid before getting pregnant and in early pregnancy increases the risk of having a pregnancy affected by neural tube defects, including anencephaly.\[^7\]

There has been a 27% decline in pregnancies affected by neural tube defects (spina bifida and anencephaly) due to fortifying grains with folic acid\[^8\]

Women taking anticonvulsant medication.
Women with undiagnosed or uncontrolled diabetes.
Women with Malabsorption problems.

**Symptoms**
The most common symptoms of anencephaly may include:
- Absence of bony covering over the back of the head
- Missing bones around the front and sides of the head
- Folding of the ear
- Cleft palate - a condition in which the roof of the child's mouth does not completely close, leaving an opening that can extend into the nasal cavity
- Congenital heart defects
- Some basic reflexes, but without the cerebrum, there can be no consciousness and the baby cannot survive

**Diagnosis**
The diagnosis of anencephaly may be made during pregnancy, or at birth by physical examination. The baby's head often appears flattened due to the abnormal brain development and missing bones of the skull. (Image 3)

Diagnostic tests performed during pregnancy to evaluate the baby for anencephaly include the following.

- Diagnosis is usually made by the AFP\(^9\) (alpha-fetoprotein) test at 16 to 18 wks and then confirmed by follow up level 2 ultrasound. Alpha-fetoprotein - a protein produced by the fetus that is excreted into the amniotic fluid called Maternal serum alpha feto protein (MSAFP), its levels are highly elevated (x 2.5 MoM) in all the neural tube defects, anencephaly usually gives the highest elevation in MSAFP. Normally, a detectable and predictable amount of this protein crosses the placenta and enters the blood stream.
alpha-fetoprotein test detects amounts of this protein, which is produced by the unborn babies' liver, and measures the amount in the mother's blood. But if the fetus has an abnormal opening in its spine (spina bifida) or head (anencephaly), or an abdominal wall defect, more alpha-fetoprotein may leak out which will cause the AFP test to be elevated.

- Amniocentesis - a test performed to determine chromosomal and genetic disorders and certain birth defects. The test involves inserting a needle through the abdominal and uterine wall into the amniotic sac to retrieve a sample of amniotic fluid.

- **Radiographic features:** Antenatal ultrasound: Anencephaly may be sonographically detectable as early as 11 weeks. It has been shown that a transvaginal ultrasound is better at diagnosing anencephaly before 16 wks than the standard abdominal ultrasound. Ultrasound can be a non invasive, cost effective and fast method to detect anencephaly and has an accuracy of approximating 100% at 14 weeks. In Ultrasound no tissue above the orbits and absent calvarium: parts of the occipital bone and mid brain may be present.(Image 4)

  ![Image 4](image4.png)

- If a small amount of neural tissue is present it is then termed exencephaly, this may be seen at an earlier stage
- Less than expected value for crown rump length (CRL) are present in Anencephaly.
- A "frog eye" or "mickey mouse" appearance may be seen when seen in the coronal plane due to absent cranial bone/brain and bulging orbits.
- Anencephaly may show evidence of polyhydramnios: from impaired swallowing

**Treatment:** There is no medical treatment for anencephaly. Due to the lack of development of the brain, approximately 75 percent of infants are stillborn and the remaining 25 percent of babies die within a few hours, days, or weeks after delivery.
Prevention

It is important for women who may become pregnant to get enough folic acid. There is good evidence that folic acid can help reduce the risk of certain birth defects, including anencephaly. Women who are pregnant or planning to become pregnant should take a multivitamin with folic acid every day. Taking 5 miligrams of folic acid daily for 2 to 3 months before conception and until at least the 3rd month of pregnancy is recommended for all woman at risk of having a child with a neural tube defect. Woman who have had a previous baby with anencephaly are 10 to 15 times more likely than average to have a second NTD pregnancy. Folic acid is thought to be able to prevent 70% of NTDS. Normal recommended daily dosage for all woman of child bearing ages is 400 micrograms (0.4 miligrams). Many foods are now fortified with folic acid to help prevent these kinds of birth defects. Genetic counselling may be recommended by the physician to discuss the risk of recurrence in a future pregnancy as well as vitamin therapy (a prescription for folic acid) that can decrease the recurrence for open neural tube defects.

Folic Acid: Folic acid is a necessary element needed for correct neural tube development. It is part of the B-Complex Vitamins and is also called folate or folacin. It is present in many leafy, green vegetables, orange juice, dried beans, and peas. It is difficult to get enough folic acid by diet alone. In fact, in order to get 4 mgs of folic acid naturally daily, we need to drink 80 glasses of orange juice or have 100 servings of broccoli. So Folic acid now fortified with many of our grain products, such as bread to help prevent neural tube defects. Folic Acid can significantly reduce the risk of having a baby with a neural tube defect.

REFERENCES


