



Anencephaly - A Case Report and Lessons for the Future

Fariha Sabeen*

Senior Resident of Anatomy, Mahatma Gandhi Memorial Medical College,
Jharkhand, India

*Corresponding Author: Fariha Sabeen, Senior Resident of Anatomy, Mahatma
Gandhi Memorial Medical College, Jharkhand, India.

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Abstract

The main objective of this particular case report is to create awareness among the people about the importance of folic acid intake, not only during pregnancy but even before it. As anencephaly could be prevented but not cured, hence a small step such as eating food rich in folic acid, along with folic acid supplementation can prevent many from this dreadful ill-fated disorder and the mothers from the emotional turmoil.

Keywords: Anencephaly; Folic Acid; Pregnancy

Introduction

Anencephaly is an embryological malformation of the central nervous system, invariably lethal, characterized by the absence of the brain and cranial vault along with other defects of the cranial structures. From the embryological perspective, anencephaly results from a failure in the neural tube closure. It is a relatively rare anomaly with incidence up to 1 in 20,000 live births [1].



Figure 1: Anencephalic female.

The newborn may be blind, deaf, unconscious and usually dies during the birth process or within a few hours. It can be diagnosed in-utero on ultrasound examination in the 11th-12th week of gestation and by elevated maternal serum levels of alpha fetoprotein (AFP) [2].

It is usually associated with polyhydramnios. About 65% of the cases of anencephaly die in-utero, and some may be delivered prematurely. It has a multifocal pattern of transmission, with the interaction of environmental and genetic factors. It has been found that folic acid supplementation reduces the risk of anencephaly to a great extent [3].

Neural crest cells originating in the neuroectoderm form the facial skeleton and most of the skull. These cells also constitute a vulnerable population. As they leave the neuroectoderm, they are often a target for teratogens. Exposure of the mother to teratogens also increases the risk of such defects [4].

Materials and Methods

A pregnant woman of G2P1, aged 28 years, presented with pain abdomen and watery discharge per vagina at 29 weeks of gestation. Her first pregnancy was uneventful with vaginal delivery at home.

On examination, her abdomen was over distended with fundal height of 32 weeks. There was no history of any ANC and Folic Acid intake. Her USG whole abdomen showed absence of skull bone. Diagnosis of anencephaly was confirmed. Mild polyhydramnios was detected. No other congenital anomaly was present.

Results

Parents were informed about the congenital disorder. Consent of termination was received from them. Labor was induced with prostaglandin E1 and a stillborn anencephalic female was delivered vaginally. The baby weighed 1.2 kg. External examination revealed complete absence of the cranium. Cerebrum and cerebellum were reduced, but the brain stem was present. The eyeballs were protruded, which gave the appearance of a frog. The remaining of the facial features and body were typical to a developing premature baby (Figure 1).

Conclusion

This case highlights the importance of regular ANCs, along with Anatomical Anomaly Scan. Anencephaly is always fatal, so any heroic maneuvers are not indicated after birth. The parents must be referred to a genetic counselor. The recurrence risk for future pregnancies is 2-5%. 70% of the defects can be prevented if women take 400mg of folic acid daily, beginning 3 months prior to conception and continuing throughout pregnancy. Because 50% of pregnancies are unplanned, it is recommended that all women of childbearing age take 400 mg of folic acid daily. If there is prior history of such defects, then it is recommended that she takes 1,000 mg per day when she plans pregnancy.

The author recommends regular ANCs with Anatomical Anomaly Scan, so that such birth defects may be diagnosed beforehand and terminated after proper counseling. In the view of the present author, food products fortified with folic acid, such as maize and wheat, should be consumed by women of child bearing age.

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