

A Case Report On Anencephaly

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Abstract

Introduction: Anencephaly is a neural tube defect which is occurred in cranial region due to the failure of neural tube closure. In India, a survey showed the incidence to be 4.5 in 1000 live births, higher than the West.

Case Report: During collection of placenta samples for Thesis works. An 18-year-old women admitted in Gynaecology Ward with chief complaints of Primigravidarum 23⁺² weeks with Anencephaly. After proper counselling with patient's husband & their legal guardian as there is no viability of this fetuses. Planned for ERPC under General Anaesthesia.

Conclusion: After studied this case, regardless of the region or country, the folic acid administration is significantly reduced the NTD rates. As well as to create awareness, it's a preventable lethal condition in our society.

Keywords: Anencephaly, Neural Tube Defect. Ultrasound, Prenatal Diagnosis, Folic Acid Deficiency.

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I. Introduction:

Anencephaly is a severe developmental defect of the central nervous system in which the developing forebrain and variable amount of the brainstem are exposed in utero, due to failure of the neural tube to close at the base of the skull in the third or fourth week (days 26-28) after conception¹. The brain and cranial bones are grossly malformed. Cerebral and cerebellar structures are reduced or grossly malformed, but the hindbrain is present. The etiology seems to be caused by genetic and environmental factors. It's prevalence at birth ranges from 1 in 5000 to 1 in 2000 which has great geographic variation².

II. Materials And Methods:

A 20-years aged of pregnant women with primigravida in 23⁺² weeks of gestation was admitted in Labor Room of GB Pant Hospital which was a referred case from Peripheral Rural Hospital for Evacuation of Retained Product of Conceptus as Anencephaly was earlier diagnosed with Ultra-sounds. No history of any chronic illness, drug or radiation exposure identified. There was no history of Folic Acid supplementation in her diet as it was an unplanned pregnancy. The postpartum period was uneventful and the patient was discharged after 72 hours of medical observation.

III. Observations:

It was seen that fetuses with Anencephaly usually diagnosed at 12-13 weeks of gestation. Ultrasound findings can be normal until the onset of ossification were not completed. A first trimester scan definitely allows a reliable diagnosis and active management of Anencephaly.

On the observation, the fetus showed absence of calveria, short neck, low set ears and eyeballs. On the Examination, CRL is measured as 23 cm (Figure 1) and weight was 300 gm. Thoracic cage was normal including the all four limbs. Cranial vault is not developed. After Dissection, these are the findings as follows-- i) Absence of Cranial vault, Frontal bone, Anterior cranial fossa, Orbital cavity, Forebrain, Hindbrain and Midbrain, ii) Presence of Brain-stem, iii) Malformed Eyeball, Occipital, Temporal bones and Middle & Posterior Cranial Fossa. Abdominal organs were normal, and there was no other associated congenital anomaly.



Fig-1: Anencephaly fetus 23⁺² weeks



Fig-2: Decapitate Head with Brain stem.

IV. Discussion:

Neural tube defects are birth defects of the before brain, spine, and spinal cord. It was seen that in the 1st month of pregnancy, often a woman even knows that she is pregnant. There were two most common neural tube defects are spina bifida and anencephaly. Among these two, anencephaly is one of the most common, the incidence of anencephaly is 1:1000- 1:2000.³ In India, a survey showed the incidence to be 4.5 in 1000 live births, higher than the West. Various genetic and environmental factors apparently account for the variability.⁴ Epidemiology studies showed that there were variation in prevalence rates. The highest rate is in Great Britain and Ireland, and the lowest is in Asia, Africa and South America. Anencephaly was seen in the 6 times more frequent in White than in Blacks, females are more often affected than males.^{5,6} It was seen that around 50% to 70% of Neural Tube Defects can be prevented by supplementing Folic Acid in early pregnancy and is encouraged regardless of the region/ country to the significant reduction. In case, Family history of such defects 4000µg per day starting 1 month before and first 3 months of pregnancy have to be continued. FA (Folic Acid) supplementation as been shown to reduce the incidence and recurrence of NTDs and possibly reduce the occurrence of several other birth defects such as cardiac, oro-facial, limb, and renal anomalies.⁷

The preventive measures include diet supplementation with folic acid before pregnancy and in the 1st month which can decrease both the frequency and severity of the condition.⁸ Another measure to be used is the fortification of both wheat and maize flour with folic acid.⁹ A secondary line of prevention is to detect the abnormality as soon as during the pregnancy, obtained by the implementation of the program of the prenatal diagnosis.¹⁰ These knowledge will be very helpful for early diagnosis and further management of the neural tube defect cases.

V. Conclusion:

Anencephaly may be diagnosed by transvaginal sonography as early as 11 weeks. All anencephalic fetuses will have an abnormally elevated maternal serum AFP. Anencephaly is a gross congenital anomaly. It is a type of neural tube defect that results from a failure of the anterior (rostral) portion of the embryonic neural tube (anterior neuropore) to close properly. The prognosis of anencephaly is very poor. Termination of pregnancy could be the best option but, it is not practiced in India due to various cultural and religious beliefs. Awareness regarding these lethal defects is very low among the population. As no curative treatment is available, So created awareness should be the key component in the prevention of such problems.

Abbreviations:

NTD-Neural Tube Defect
FA-Folic Acid
CRL-Crown Rump Length
USG-Ultra sonography/ Sounds
AFP-Alpha fetoprotein

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Declaration:

The study was approved by Institutional Approval Board and Institutional Ethical Committee.

Conflict Of Interest:

The authors declare no conflict of interest.

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