

## Case Report

# MEROENCEPHALY

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## ABSTRACT

Anencephaly which is basically a misnomer used in place of meroencephaly for a long time is one of the most common birth defect that is seen in stillborn fetuses. It has multifactorial relations with environment, genetics as well as nutrition. It can be diagnosed by ultrasound, serum alfafetoprotein (AFP) level. The present study was done of a female aborted fetus of 32 weeks having anencephaly whose specimen was present in our department. So we planned to present a case report of this very anomaly with its development and genetic causes that lead to this lethal but preventable congenital defect.

**KEYWORDS:** neural tube defect, anencephaly, meroencephaly

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## INTRODUCTION

Neural tube defects are amongst the most common congenital anomalies. Meroencephaly (partial absence of the brain) is the most severe neural tube defect and is also the most common anomaly affecting the central nervous system. The term anencephaly which is commonly used is a misnomer because a remnant of the brain tissue is present [1]. It's a type of upper neural tube defect that results in failure of the brain to develop [2]. Polyhydramnios may be associated with meroencephaly (anencephaly). It is also associated with acrania that is complete or partial absence of the neurocranium. Seventy percent of the defects can be prevented if women take 400micrograms of folic acid daily beginning prior to conception and continuing throughout pregnancy. If a woman has had a child with neural tube defect or if there is a history of such defects in her family, it is recommended that she takes 400 micrograms of folic

acid daily and then 1000 micrograms per day when she tries to become pregnant [3].

## Case Report

The study was carried out on an aborted female fetus of 30weeks. There was a history of polyhydramnios and no history of folic acid supplementation. The fetus showed absence of a major portion of scalp and cranial vault and the defect extended up to the cervical vertebrae. The brain tissue and spinal cord in the cervical region were exposed to the outside. Below the level of cervical region no defect was observed and vertebral column was covered by normal but hairy skin because of prematurity. Nose was broad and ears were folded. The eyes were developed but the eyelids were looking swollen. As such no abnormality of lips or palate was observed. The neck with trunk was short and shoulders were broad. No other associated external deformities were observed.

**Fig. 1:** Anterior view of the aborted fetus with meroencephaly.



**Fig. 2:** Posterior view of the same fetus.



**Fig. 3:** Lateral view of the fetus. Kindly note the bulging eyeball, malformed auricle, remnant of brain tissue and poorly developed cranial vault.



## DISCUSSION

Meroencephaly is a severe defect of the calvaria and brain that resulting from failure of the rostral neuropore to close during the fourth week. As a result, the forebrain, midbrain, most of the hindbrain and the calvaria are absent. Most of the embryo's brain is exposed or extruding from the cranium—exencephaly. Because of the abnormal structure and vascularisation of the embryonic exencephalic brain, the nervous tissue undergoes degeneration. The remains of the brain appear as a spongy, vascular mass consisting of hindbrain structures. It is a lethal defect occurring in 1 in 1000 births. 2-4 times more common in females and always associated with acrania and may be associated with rachischisis when defective neural tube closure is extensive. It can be easily diagnosed by ultrasound, MRI, fetoscopy and radiography. It usually has a multifactorial mode of inheritance<sup>1</sup>. The neural tube develops and closes during the 3rd & 4th weeks after conception and is normally completed by 28 days post conception [4]. Exposure to valproic acid and other antimetabolites of folic acid and other toxins like lead etc. during critical period i.e. up to 6 weeks after last menstrual period, interfere with normal folate metabolism and increases the likelihood of anencephaly [5]. Recently, a hypothesis of "multiple site of neural tube fusion" has been investigated in animal models and in humans. Four sites of neural tube fusion have been identified. Site 1 initiates in the future cervical region between the third and fourth somites at the caudal part of the hindbrain, and progresses both caudally and rostrally. Caudally, it proceeds all the way down to the end of the neural groove until the caudal neuropore. The next two sites of initiation of fusion are located rostral to site 1. A second fusion initiates at the prosencephalon-mesencephalon boundary (Site 2) and extends both rostrally and caudally. This second fusion completely closes the roof of the telencephalon and the metencephalon. A third fusion site (site 3) progresses caudally, and closes the rostral end of the neural plate. Finally, the fourth fusion site (site 4) appears at the caudal end of the neural plate and extends rostrally to meet the fusion extending back from site 1 [6]. Phenotype of NTD will vary depending on the

involvement of the site of fusion. Van Allen et al compared the multisite model and the traditional single-site model of neural tube closure for the best explanation for NTDs in humans. With the multi-site neural tube closure model majority of NTDs can be explained by failure of fusion of one of the closures or their contiguous neuropores. They hypothesize that anencephaly results from failure of closure at site 2 and craniorachischisis results from failure of closures at sites 2, 4, and 1 [7]. The exposed base of the skull is covered only by a vascular membrane [8,9,10]. This defect can be diagnosed during alpha-fetoprotein (AFP) screening. Fetal ultrasound can also be useful for screening of neural tube defects [11]. The recurrence risk is 1.9% for parents who had one affected child. Folic acid supplementation has been shown to be an effective means of lowering recurrence risks for future pregnancies [12].

## CONCLUSION

Meroencephaly is a preventable anomaly like any other neural tube defect and it can be prevented by taking folic acid supplementation during reproductive age. Screening tests like AFP and USG can diagnose the condition early and termination of pregnancy can be done as early as possible. All the parents who had or not any previous history of this lethal defect should be educated about its modes of preventions. If possible, genetic counseling in such cases should be done.

**Conflicts of Interests: None**

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