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Case Report

# Anencephalic fetus with craniospinal rachischisis - Case report

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ABSTRACT

Anencephaly is a severe neural tube defect (NTD) caused by failure of closure in the cranial neuropore during fourth week of pregnancy. As a result, major portion of the brain, skull and scalp is absent. Anencephaly may be associated with rachischisis, where defective neural tube closure is extensive and spinal cord is exposed. Overall incidence of anencephaly is one in every 1000 births. It can be easily diagnosed by ultrasonography. Anencephaly newborns are not viable nor treatable and classified as lethal NTDs. Nutritional and environmental factors play a role in production of NTDs. Here we report and discuss a rare case of anencephalic fetus with craniospinal rachischisis of 25 weeks of gestation and their embryological origin.



## 1. Introduction

Anencephaly is a congenital severe lethal neural tube defect (NTD) occurring one in every 1000 births. It is also most common anomaly affecting the central nervous system (CNS). NTDs involve neural and non-neural tissues like vertebrae, muscles, and skin. NTDs include anencephaly (partial or total absence of brain calvaria), spina bifida, encephalocele (herniation of brain and meninges), craniorachischisis (anencephaly with abnormal vertebrae) and iniencephaly (defect in occipital region with retroflexion of neck and trunk). [1,2]

Neurulation is a process where neural plate forms neural tube that gives rise to primitive CNS. Neural plate is derivative of neuroectoderm. At the end of third week of pregnancy lateral edges of neural plate elevates and forms neural folds. Later neural folds begin to fuse in the midline, extends cranially and caudally thus forming neural tube. Cranial neuropore closes approximately on $25^{th}$ day and caudal neuropore on $28^{th}$ day thus resulting a closed tubular structure of primordial CNS. Cranial part of neural tube forms brain and caudal part develops into spinal cord. NTDs result from abnormal closure of neural folds in third and fourth week of development. [1,3]

Anencephaly is caused by failure of closure in the cranial neuropore during fourth week of pregnancy. As a result, major portion of brain is abnormal and development of the calvaria is defective. Most of the nervous tissue is exposed or extruding from skull and undergoes degeneration or atrophy. Generally this condition is referred as anencephaly (without brain), but rudimentary neural tissue is always present, hence for this reason, meroencephaly (partial absence of brain tissue) is a better term. Anencephaly is associated with acrania (absence of calvaria or skullcap) and rarely appears with rachischisis when defective closure is extensive and whole spinal cord is exposed. Rachischisis affects axial structures as a result of faulty induction by notochord or from teratogenic agents. Anencephaly newborns are not viable nor treatable. [1,3]

## 2. Case Report

A 24 year old lady with third pregnancy diagnosed with a fetus having anencephaly with craniospinal rachischisis was admitted for medical termination of pregnancy to KLEs

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Dr Prabhakar Kore Charitable Hospital &MRC, Belagavi. She belongs to low socioeconomic group, working as a labour in building construction. She had second- degree consanguineous marriage of 4 years. She did not have any habits like tobacco, pan-gutaka, and alcohol.

She was third gravida of 25 weeks and 2 days of gestation as per her last menstrual period (LMP). She had one male child with full term normal delivery of 3 years old, alive, and healthy. History of one abortion at 12 weeks of gestation was noted. Her menstrual cycle was regular with normal flow. No history of pregnancy induced hypertension, gestational diabetes mellitus, diabetes mellitus, cardiac and renal disease. She was taking iron and folic acid (IFA) tablets since beginning of her pregnancy. No family history of NTD occurrence and claimed that she had not used any medication other than IFA.

On examination, she was moderately built, nourished with 150cm in height and 46kg in weight. Her pulse, heart beat and blood pressure was normal. Random blood glucose was 80mg/dl. Urine, HIV and HbSAg tests showed negative result. As per ultrasonography (USG) she had single intrauterine gestation with cephalic presentation of 25 weeks of gestation age as per LMP with estimated fetal weight of 600gms. Fetal anatomy revealed absence of cranial vault, and spinal defect in lumbar region measuring 2.0 x1.7cm and reported as anencephalic fetus with craniospinal rachischisis.

She delivered male macerated stillborn baby of 300 gm by vaginal route. On examination of fetus, scalp and calvaria were absent and brain was exposed with protruded eyes (Figure 1 A). Brain was maldeveloped and replaced by angiomatous mass. Skin was absent over spine exposing whole spinal cord (Figure 1 B). Diagnosed as an anencephalic fetus with craniospinal rachischisis. Umbilical cord was normal showing one vein and two arteries. Genetic study was not carried out.


**Fig. 1:** A & B – showing anencephaly fetus with craniospinal rachischisis

## 3. Discussion

Prevalence of anencephaly varies from country, race, sex and environmental factors. In a population based study in India frequency of NTDs ranging from 6.57-8.21 per 1000 births, in that anencephaly was reported in 2.5 in 1000 births.[4] In another prospective study of 3500 consecutive births, in south India 11.4/1000 births of NTDs were found, in that 5.1/1000 births related to anencephaly with craniospinal rachischisis. In previous history of NTDs and in consanguineous marriage cases showed increased risk of having NTDs were more than others.[5] Highest 11.39/1000 live births incidence of NTDs were found in china[6] whereas lowest 1/1000 live births incidence was reported in USA.[7]

In developing country, large number of congenital malformation and genetic disorders are one of the causes for infant mortality and morbidity. A study carried on 94640 newborns to know the prevalence of malformations showed 2.03% rate of malformed babies and commonest are NTDs and musculoskeletal disorders.[8]

NTDs are believed to originate from complex interaction of environmental and genetic factors. Environmental factors like age, periconceptual infection, recreational drug use, caffeine, smoking and alcohol influence the genesis of NTDs.[3,7,8]

Extensive clinical and epidemiological research has demonstrated that poor maternal nutrition mainly folic acid seen in low socioeconomic group increases the risk of NTDs. Other micronutrients like vitamin B6, B12 and minerals like zinc are also important for proper development on neural tube. Certain drugs like, valporic acid, anticonvulsant and exposure to high levels of vitamin A produces NTDs.[9–11] Low maternal vitamin B12 increases the risk of NTDs. Measurement of holotranscobalmin (holo TC) is a sensitive indicator of vitamin B12 status.[9]

Genes involved in folate metabolism are believed to be important in production of NTDs. Gene like Methylene Tetra Hydro Folate Reductase (MTHFR) mutation is responsible for folate related NTDs.[12,13] Administration of food fortified with folic acid (400$\mu$g) and synthetic vitamin B12 during periconceptual period has reduced the 50-70% risk of NTDs in USA.[12,14]

To conclude NTDs are strongly suspected in utero when there is high level of alpha feto protein (AFP) in maternal serum and amniotic fluid. Therefore, in risk pregnancies, measure of AFP level and early USG may help in early identification of NTDs and genetic counseling is beneficial for future planning. Since there are no curative modalities available hence, clinical focus is mainly based on preventive measures. Studies have proven that occurrence of NTDs can be reduced by taking folic acid daily during pregnancy. Since most of the pregnancies are unplanned even an dministration of multivitamin containing 400$\mu$g folic acid should be recommended to all women of child bearing age will diminish the chances of NTDs.

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## 6. Conflict of Interest

None.

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