ORIGINAL RESEARCH PAPER

Radiology

KEY WORDS: neural tube defect, totalis, craniorachischisis, spina bifida, congenital

# CRANIORACHICHIASIS A RARE CASE REPORT.

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**ABSTRACT**

Craniorachichiasis is an extreme example of defective neural groove closure. It is a major type of neural tube defect that refers to congenital fissure in the skull and vertebral column. It results when the neural tube fails to close during the third and fourth weeks of gestation. We present a case of craniorachichiasis in a female fetus. The defect in skull vault is seen extending upto the lower thoracic part of the vertebral column. The brain tissue and spinal cord were exposed to the exterior. The neck was short.

## INTRODUCTION:

Neural Tube Defects are congenital malformations which arise from incomplete closure of neural tube during early embryogenesis. It occurs between 17 to 30 days of gestation at the time when the mother may not be aware that she is pregnant and the embryo is estimated to be about the size of a grain of rice. Incidence rate ranges from 1.2 to 1.7 per 1000 births. Most common type of neural tube defect found in human is spina bifida and anencephaly. Craniorachischisis Totalis [Total Dysraphism] is a rare form of neural tube defects in which brain and spinal cord are exposed. This defect usually results in early spontaneous abortion often associated with malformation of other organ system. Formation of neural tube first occurs on day 22 at the level of somite 3; fusion preceding both rostrally and caudally and the neural tube is closed by the day 26. Primary disturbance in the axial mesoderm system may impair formation, elevation and approximation of neural folds leading to craniorachischisis totalis.[1] A multifactorial inheritance is considered to be responsible along with environmental factors like nutrition and folic acid, chromosomal aberration and some teratogens. A case of Craniorachischisis Totalis from our district hospital has been dissected, studied and the findings, associated malformations have been discussed in this case report. Likelihood of having child with neural tube defect increases significantly once one affected offspring is born. It is recommended that Folic acid substitution during the period beginning two months prior to conception and continuing through gestation reduces incidence of neural tube defect by as much as 70%. Hence early detection of the anomaly and termination of the pregnancy followed by counseling of the parents will help to prevent the further occurrence of the defect in the subsequent pregnancies.[2]

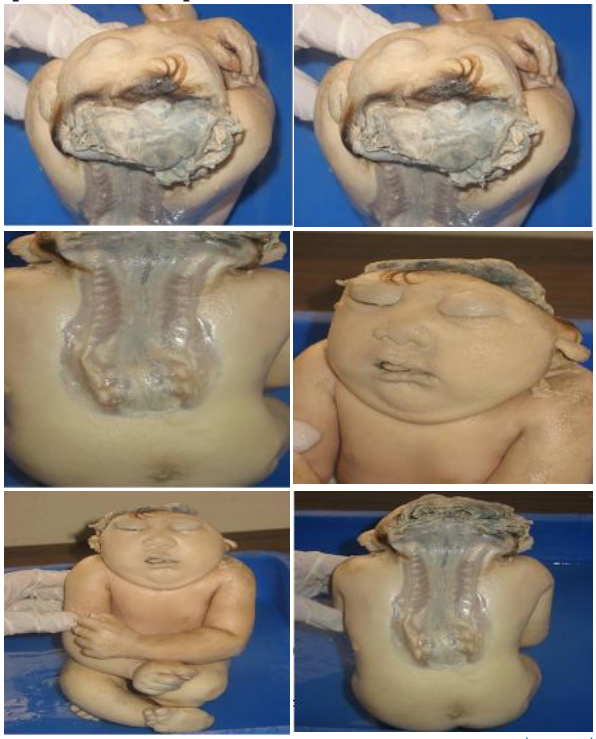
Craniorachischisis is the most severe form of neural tube defect in which both the brain and spinal cord remain open to varying degrees. It is a very rare congenital malformation of the central nervous system. The prevalence is not known. Craniorachischisis totalis, the most complete form of craniorachischisis, presents anencephaly and total spina bifida together and is lethal. As with other neural tube defects, craniorachischisis is thought to be of multifactorial origin. Antenatal diagnosis is possible by ultrasonographic monitoring.[3]

These cases have mutations in both CELSR1 and SCRIB. Functional effects of these unique or rare human variants were evaluated using known protein–protein interactions as well as subcellular protein localization.[4]

The incidence of anencephaly and spina bifida is usually higher in groups with lower socioeconomic status and at least half the cases of neural-tube defects can be prevented if women consumed sufficient amounts of the folic acid before conception and during early pregnancy. The fetus was of 13 weeks of gestational age, the crown rump length measured 118 mms. The fetus had anencephaly and spina bifida , the brain and calvaria were absent. In addition to these anomalies, it had exomphalos and sternal cleft. The fetus went for spontaneous abortion at a rural hospital of Kerala state in South India. The detailed obstetric history of the mother was not available.[5]

## Case Report:

We present a case of craniorachichiasis in a 28 weeks female fetus. The defect in skull vault is seen extending upto the lower thoracic part of the vertebral column. The brain tissue and spinal cord were exposed to the exterior. The neck was short.



## DISCUSSION:

Early detection - alpha fetoprotein screening and ultrasound scan.

There is no cure or standard treatment. Infants born with craniorachischisis die within hours of birth. Therefore, pregnancy should be terminated if diagnosed early.

Can be prevented by taking folic acid supplementation during reproductive age.

## CONCLUSION:

- Rare but severe congenital malformation.
- Combination of anencephaly and non-closure of the spinal cord.
- Prevented by folic acid supplementation

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