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Original Article

A cadaveric foetal study of craniorachicchisis totalis

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ABSTRACT

Aim of the study is to study in detail about Neural tube defects and the importance of folic acid during antenatal period. Neurulation is the process whereby the neural plate forms the neural tube. Cranial end is called anterior neuropore and caudal end is called Posterior neuropore. We received a male dead fetus from Institute of obstetrics and Gynaecology, Govt Rajaji Hospital Madurai. External examination of fetus showing Craniorachischisis was observed. Detailed study of fetus was performed. Craniorachischisis is a rare and most severe form of Neural tube defect in which both Brain and spinal cord remain open. Both Anencephaly and Spina bifida are present. Craniorachischisis can be diagnosed prenatally by ultrasound and by determination of Alpha fetoprotein (AFP) levels in maternal serum and amniotic fluid. It is estimated that 50-70% of Neural tube defects can be prevented if women take 400ug of Folic acid daily at least 1 month prior to conception and continuing throughout early pregnancy.

Keywords: Craniorachischisis, Ancencephaly, Spina Bifida, Neural tube defect (NTD)

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INTRODUCTION

Craniorachichisis is a rare and most severe form of Neural tube defect in which both Brain and spinal cord remain exposed .Both Anencephaly and spina bifida are present. Neural tube defects occur when the neural tube fail to close. If the neural tube fail to close in the cranial region, then most of the brain fail to develop .There is failure in the development of vault of the skull. This defect is called anencephaly. If the closure fails caudally anywhere from the cervical region to sacral region, then the defect is called Spina bifida.

Most common site for Spina bifida to occur is in the lumbosacral region. If the entire length of the neural tube remains open from the surface of the head on to the back, then the defect is called craniorachichisis totalis [1]. Miscarriage of the fetus commonly occurs during pregnancy .Usually fetus dies shortly after birth or stillbirth occurs. Some cases of craniorachichisis may be associated with other anomalies like Omphalocele, Congenital diaphragmatic hernia, Renal anomalies and Cardiac anomalies.[2,3]

Origin of Neural tube defect is multifactorial and the likelihood of having a child with such defect increases significantly once one affected offspring is born. Folic acid deficiency[1,6] plays an important role in occurrence of Neural tube defect. Incidence of Neural tube defects involving Brain or Spina cord or both is 0.5-2 per 1000 pregnancies worldwide. Incidence of neural tube defects in India ranges from 0.6-13/1000 live births and varies in different populations[4]. More common in females than males. Polymorphism in the Methylenetetrahydrofolate reductase (MTHFR) gene can be a susceptibility factor in some population. Reduction in MTHFR activity results in decreased plasma folate levels[5] .Recently, mutations in the VANGL genes have been identified and associated with neural tube defects. Neural tube defects show an incidence of 5-10% in Trisomy 13 and Trisomy 18[1].Poor socioeconomic status, Multiparity, Maternal hyperthermia, Valproic acid exposure, Hypervitaminosis[5]

M ATERIALS AND METHODS

Of the ten foetuses received from Institute of Obstetrics and Gynaecology, Govt Rajaji Hospital Madurai during the year 2017 , one dead male fetus of 28weeks gestation having craniorachichisis totalis with anencephaly and spina bifida was observed. Academic autopsy was performed in Institute of Anatomy to see the presence of other associated congenital malformations. Their findings were appropriately documented.

OBSERVATION

The fetus on external examination showed craniorachichisis .It showed absence of scalp ,cranial vault. The defect extended upto lower thoracic region. Brain tissue and spinal cord in the cervical and lower thoracic region were exposed.Below the level of lower thoracic region no defect was observed and vertebral column was covered by normal skin. Eyes were bulging and there was absence of neck .Protruded tongue and prominent ears were observed. Fetus had extreme extension of head with Frog like appearance of face. Fetus had imperforate anus also .No other abnormality was found externally. Academic autopsy was carried out and no abnormalities were found internally.

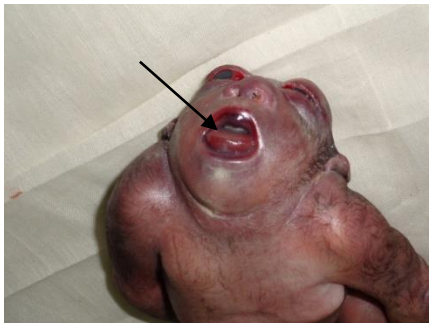

Fig 1: Protruded tongue and absence of neck

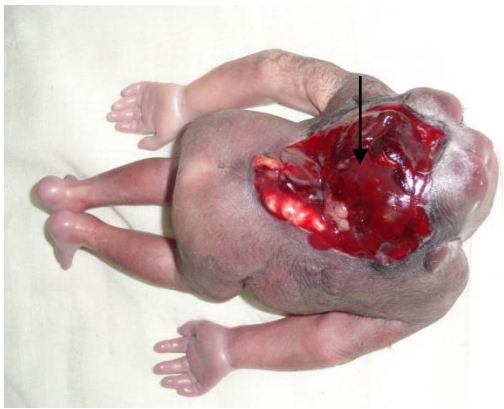

Fig 2: Exposed Brain tissue and spinal cord

Fig 3:Prominent ears

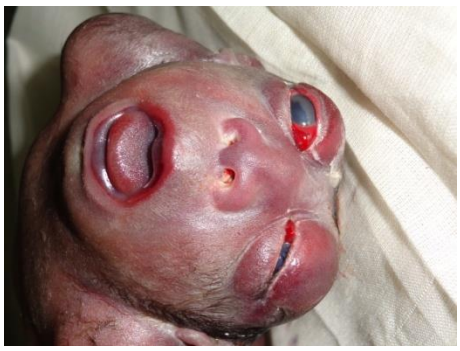

Fig 4:Frog like appearance of face

DISCUSSION

Neurulation is the process whereby the neural plate forms the neural tube. By the end of the 3rd week, the lateral edges of the neural plate become elevated to form neural folds, and the midregion forms the neural groove. Gradually the neural folds approach each other in the midline, where they fuse. Fusion begins in the cervical region and proceeds cranially and caudally. As a result, the neural tube is formed. Cranial end is called anterior neuropore and caudal end is called posterior neuropore. Neural tube communicates with the neural cavity through ant and post neuropore. Closure of the cranial neuropore occurs at 25th day and posterior neuropore at 28th day.Hence CNS is represented by a closed tubular structure with a narrow caudal portion, the spinal cord and a broader cephalic portion characterized by a number of dilatations called Brain vesicles. Most defects of the spinal cord result from abnormal closure of neural folds in the 3rd and 4th weeks of development resulting in abnormalities involving meninges, vertebrae, muscles and skin. Recent evidence proved that Folic acid reduces the occurrence of Neural Tube Defects by as much as 50-70% if 400ug is taken daily atleast one month prior to conception and continued throughout early pregnancy.[1,6]

Dr.Jose Ignacio (1992), reported a case of craniorachichisis associated with sirenomelia and hypoplasia of phlanges of right thumb. Ashutosh Halder, (2001) reported a case of sirenomelia associated with craniorachichisis.Associated cardiac abnormality (primitive heart), Renal agenesis, absent external genetalia were also observed. Kiran G (2009) reported a case of craniorachisis totalis with only external abnormalities and they found no abnormalities internally. NS Naveen, (2010), observed a 13 weeks old female fetus with Craniorachisis .sternal cleft and examphalos were also seen with cranioniorachichisis, N.Ratnakar rao (2015) reported a case of craniorachichisis with only external abnormalities.

Singh A, (2016) observed a 19 weeks old male fetus with congenital diaphragmatic hernia associated with craniorachichisis during 2017 we observed 10 fetuses ranging from 28-30 weeks. One fetus was observed with the following deformities, Frog like appearance of face, Protruded tongue, Protruding eyes with absence of neck, Absence of scalp and cranial vault, Deformed ears and Anal atresia. The incidence of craniorachichisis observed by us from our study was 0.5 %.

CONCLUSION

Craniorachichisis can be diagnosed prenatally by ultrasound and by determination of AFP levels in maternal serum and amniotic fluid[6]. Vertebral column can be visualized by 12 weeks of gestation and the defect in closure of the vertebral arches can be detected. The prognosis of craniorachichisis is exceptionally poor and death of the neonate is unavoidable.

Experimental treatment is available only for spina bifida.It involves to perform surgery in uterus as early as 22 weeks gestation. The fetus is exposed by an incision into the uterus and the defect is repaired and infant is placed back in the uterus.[1]
In this case the mother belongs to low socioeconomic status and there was a history of non-consumption of Folicacid during antenatal period which could be the etiological factor for craniorachichisis. It's estimated that 50-70% of NTDs can be prevented if women take 400ug of Folic acid daily atleast 1 month prior to conception and continuing through early pregnancy.50% of the pregnancies are unplanned so it is recommented that all women of child bearing age should take a multivitamin containing 400ug of folic acid daily.[1,6]

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