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MAJOR NEURAL TUBE DEFECTS WITH FACIAL DISMORPHISM- A CASE REPORT

Anatomy

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ABSTRACT

Neural tube defects [NTDs] are the disorders caused due to non closure of neural tube during 3rd to 4th week of embryonic period. NTDs have an incidence of 1-2 per 1000 births. Major types of NTDs include anencephaly, spinabifida, craniorachischisis, encephalocele, inencephaly. Present study was carried out to study craniorachischisis and associated malformations. Method: study was carried out on 24 weeks aborted female fetus showing gross neural tube defect, in department of anatomy. Result: fetus was having craniorachischisis i.e. complete failure of neurulation along with facial dismorphism and other malformations.

KEYWORDS

neural tube defects, craniorachischisis.

INTRODUCTION

Neural tube defects [NTDs] are the disorders caused due to non closure of neural tube during 3rd to 4th wk of embryonic period. Amongst severe congenital malformations, NTDs are more common having incidence of 0.5-2/1000 pregnancies worldwide1.Most of them are lost as a result of spontaneous or elective abortions[2].

Formation of brain and spinal cord begins with development of neural tube, through the embryonic process of neurulation. Failure of initiation of closure at different sites leads to varies types of NTDs. Craniorachischisis a severe form of NTD, is due to failure of initiation of closure at closure-1 site[1].

Craniorachischisis is an example of defective neural groove closure, combining anencephaly and total spina bifida with meningomyelocele. Open and close are the two types of NTDs which can be classified according to coverings of defect. NTD is open, when neural tissue is exposed to exterior or covered only by membranes and close, when defect is covered by normal skin and not exposed to exterior[2].

We report a case of craniorachischisis which is severe form of open NTD, and was associated with other malformations.

MATERIAL METHOD

The study was carried out in department of anatomy, MGM Medical College, Aurangabad. Fetal specimen was obtained from dept of OBGY, and after taking ethics clearance from institutional ethics committee, examination was carried out.

On external examination, fetus was studied for defects in cranium, vertebral column, face and other skeletal deformities. Dissection was carried out by taking midline incision on chest and anterior abdominal wall. Internal examination was done to find out any other internal systemic anomalies.

CASE REPORT

A 23 yr old primigravida from low socioeconomic status came with fetal ultra sound report suggesting of 24 to 26 weeks gestation with NTD and no fetal cardiac activity. She delivered still born female baby. Fetal specimen was of 24 weeks as confirmed by 14.85cm crown rump length. Weight was 862gm.

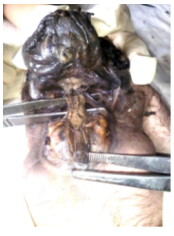

Figure 1

CRANIUM AND VERTEBRAL DEFECTS–

Fetus showed absence of major portion of scalp and cranial vault and defect was extending to the lower thoracic region. Degenerated brain and spinal cord was covered only by membranous tissue, lacking skin over it. Spinal cord and spinal nerve rootlets were seen exposed to exterior. [As shown in image 1]

Incomplete closure of vertebral arches with hyperextension of malformed cervico thoracic spine was seen.

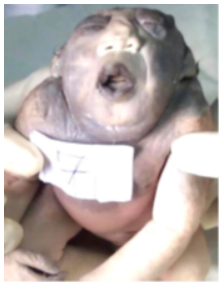

Figure 2

FACIAL DEFECTS–

face was upwardly turned, ears folded, nose was broad. Mandibular skin directly continuous with that of chest owing to the lack of neck. [see image 2]
No cleft lip but posterior cleft palate was present.

CARDIOPULMONARY DEFECTS-

On internal examination, heart appeared normal. Hypoplasia of lungs seen. Right lung was having only two lobes separated by single fissure with reduced weight. The weight of left lung was also low, but with normal morphology of lobes and fissure. [see image 3]

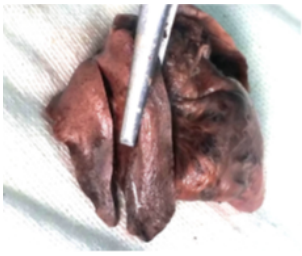

Figure 3

ABDOMINAL DEFECTS-

Both adrenals were apparently absent. Kidneys and other systems were normal.

DISCUSSION:

NTDs are the most severe and frequently encountered congenital

anomalies.

Anencephaly is absence of most of the brain and cranial vault, and if defect extends beyond cranium then it is called as craniorachischisis.[3]

The neural tube develops and closes by the process of neurulation. During 3rd to 4th week of gestation and is normally completed by 28 days post conception.[4]

According to copp et al study, in humans, Initiation of closure of neural tube occurs at different sites. And failure of closure initiation at different sites leads to various types of NTDs. Failure of closure initiation at Closure-1 site leads to craniorachischisis; which includes open neural tube in the midbrain, hindbrain and whole of the spinal region. Whereas incomplete closure at cranial neuropore causes anencephaly and failure of posterior neuropore closure leads to open spina bifida[1].

Craniorachischisis totalis represents complete failure of neurulation, where neural plate like structure is present, but no overlying axial skeletal or dermal coverings form. So, brain and spinal cord remain open to varying degrees[1]. In this case we found, total anencephaly along with open bifid spine covered only by membranes. Whereas in close spina bifida, defective vertebral arches and neural tissue are covered by skin.

According to study of R. Deopujari, et al, craniorachischisis presents with vertebral and cranial vault defects, bulging eyes, folded ears and broad nose.

In present study along with above findings, we observed that face was upturned and neck was absent, head seems to arise directly from chest. Anencephaly and other neural tube defects may also present with short thorax, clubfeet, a large thymus, pulmonary hypoplasia, flattened pituitary fossa, hypoplasia of adrenal cortex, cleft palate and renal defects.

In present study, pulmonary hypoplasia was present along with bilateral absent adrenals, fetus also showed posterior cleft palate.
The study done by stoll et al suggested that, 'presence of associated malformations with NTDs depends upon type and level of NTD and by severity of dysraphic defects. And that the frequency of associated malformations is more with severely dysraphic defect, craniorachischisis and encephalocele. 'They found that renal malformations and facial cleft are more frequent associated malformations followed by skeletal malformations[2].

In NTDs, because of lack of calvaria and vertebrae, open neural tube is exposed to the amniotic fluid insult; along with mechanical trauma, this leads to massive loss of neural tissue by the end of pregnancy. With upcoming advanced surgical procedures we can cover this neural tissue if defect is small, during intra uterine life only[1].

The aetiology of NTD is multifactorial. Both environmental and genetic factors are may be responsible for causation of NTDs. Most accepted environmental factor include deficiency of folate both before and during pregnancy [1,3,5]. and thus many studies support that folic acid supplementation in periconceptional period decreases the birth prevalence of NTDs[5]. Although the fact how the folic acid prevent NTD is under study. Other environmental causes include deficiency of inositol, vit B12, zinc. Exposure to valproic acid and folate antagonists. Glycemic disregulation like diabetes malietes and thermal disregulation also plays a role[3].

Genetically NTDs can be associated with different known genetic syndromes[like Meckel syndrome, anterior sacral meningocele, anal stenosis,etc], trisomy18, trisomy13, triploidy and various chromosome rearrangement[3,5,6]. In study of stoll et al, 32% of infants with recognizable conditions had chromosomal abnormilities amongst which most of them had trisomy18[5].

Polyhydroamnios during pregnancy is seen in majority of cases due to lack of swallowing mechanism. Fetal ultrasound and raised alpha feto protein levels can be used for screening of NTDs.

Prognosis is exceptionally poor, there is no cure or standard treatment available for craniorachischisis till date. Most of the cases are still born or spontaneously aborted. If born, death of neonate is unavoidable. There for if diagnosed early, termination can be decided earliest possible. Genetic counseling of parents may be helpful in this aspect.

Recurrence risk for siblings of index cases is 2-5%, with further increase in risk after 2 or more affected pregnancies. This can be reduced after folic acid supplementation.

CONCLUSION

NTDs are common congenital anomalies, considering the increased incidence of it, the study of associated anomalies and knowledge of it may be important for academic, surgical as well as radiological procedures. Accurate and early diagnosis of NTDs is helpful in counseling of parents. Educational programs, genetic counseling and folic acid supplementation during periconceptional period will be helpful for primary prevention of NTDs.

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