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Neural Tube Defects with Exencephaly in Human Fetus

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Abstract

Background: Anencephaly and spina bifida are the commonest type of Neural tube defect. Exencephaly is the severe form of anencephaly in whichthere is not only deficiency of scalp and cranial vault but also uncovered immature braintissue protrudes out of the cranium. There isloss of fetal brain tissue to variable degrees and it is considered a precursor to anencephaly . Most cases are stillborn. Present study was carried outtostudy exencephaly and the associated morphological abnormalities. This study also compares the findings with other studies and defines a clinical entity that is incompatible with human life. *Method*: The study was carried out on a collection of 20 aborted fetuses in department of anatomy out of which 11 showed presence of exencephaly. *Result*: These exencephalic fetuses presented with facial abnormalities, cvs abnormalities, skeletal deformities and abdominal abnormalities.This study describes in detail the severe form of NTD called exencephaly and the associated abnormalities. We have described a lesser defined condition that is exencephaly, the anatomical knowledge of such may be important for academic, clinical as well as radiological procedures .With the detailed knowledge of the condition the diagnosis ofexencephaly, can be established sonographically even in the first trimester and necessary measures can be taken.

Keywords: Anencephaly; Exencephaly; Neural Tube Defect; Spina Bifida.

Introduction

The vast majority of congenital anomalies appear during embryonic period of prenatal life. Neural tube defects are the most frequent and devastating congenital anomalies during this period. Neural tube defects are malformations secondary to abnormal neural tube closure that occur between third to fourth week of gestational age [1]. Two types of neural tube defects which are commonly seen are anencephaly and spina bifida. Anencephaly is a congenital absence of a major portion of the brain, skull, and scalp [2]. The primary abnormality is failure of cranial neurulation, the embryologic process that separates the precursors of the forebrain from amniotic fluid [3]. A slightly more severe form of neural tube defect, which is discussed in detail in this article, is exencephaly. In anencephalic fetus, when there is primitive osseous defect of cranial bones the cranial bones are not formed completely and the cranial dome is not closed due to which the brain mass lacks developmental boundaries and protrudes to the exterior through the defect [4]. In this abnormality, prolonged exposure of the developing encephalon to amniotic fluid and repeated mechanical and chemical trauma in utero result in anencephaly[5, 6]. Some studies support the theory that exencephaly is the forerunner of anencephaly[7]. Many imaging studies like the ultrasonography has stated that the Cranial malformations were the most common structural fetal abnormalities in NTDs [8]. Some ultrasonographic studies has described the appearances of exencephaly in the first trimester as the 'Mickey Mouse face'[9,10]. The literature on NTDs is extensive, but few studies provide information on exencephaly and the associated abnormalities. Here we studied exencephaly and the associated abnormalities.

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Aims and Objective

The aim of this study was to describe a rare form of anencephaly that is exencephaly and study its associated anomalies in detail. The study also compared the findings with the other studies to find its clinical significance.

Material and Methods

The study was conducted in Municipal Medical College and general hospital. 20 anencephalic fetus were dissected in the Anatomy department of the institute. The fetal specimens were obtained from department of obstetrics and gynaecology from the same institute. The cases were originated from still birth, spontaneous abortion and therapeutic abortion. No parental history was available. The age was estimated by the criteria like crown rump length, development of limbs and external genitalia. The findings were done by external examination, photography and internal examination. For the internal examination a midline incision was taken on the chest and anterior abdominal wall and the walls were retracted laterally. The presence of exencephaly was recorded and any associated abnormality was studied. The fetuses were observed for the defect in the

a. cranium
b. vertebral column,
c. face and skeletal deformities

Dissection of fetuses was carried out to find out any other internal anomalies cardiovascular system, genitourinary system and abdominal system. Ethics clearance was taken from institutional ethics committee.

Results

In the present study 20 anancephalic fetuses of gestational age 16 to 30 weeks were studied. On external examination it was found that 11 fetuses were exencephalic which showed the recognizable neural tissue found outside the cranial vault (Figures 1, 2). These fetuses ranged in age between gestational age 19 and 30 weeks, and had crown-rump length between 14 and 25 cm whereas in same age group normal fetuses it is found to be 15 to 36 cm [11].

Facial Defects

In 8 fetuses the cranial vault (bony calvarium) is symmetrically absent. All the 11 fetuses showed frog-like appearance with prominent bulging eyeballs, folded ears and absent neck (Figures 1, 2). 6 cases of protruded tongue. 4 fetuses showed cleft lip and cleft palate (Figure 1).

Spina Bifida

Spina bifida was seen in 4 fetuses (Figures 3, 4).Out of the 4 spina bifida, 3 fetuses showed craniorachichisis (Figures 3, 4). The defect in this case was extending beyond the cranial vault (Figure 3). Brain tissue and spinal cord were exposed to the exterior.1 fetus showed spina bifida in lumbar region.

Cardiovascular Defects

When the cardiovascular examination was done 2 cases of VSD (Figure 6) and 3 cases of ASD (Figure 5) was found.

Abdominal and Skeletal Defects

In abdominal examination 1 case each of gastrochisis, omphalocele and umbilical hernia was found. In skeletal system 4 cases of club foot and club hand were found (Figure 7). There was 1 case of kyphosis (Figure 3). In genitourinary examination 1 case of hypospadias was seen.

Fig. 1: Fetus with exencephaly

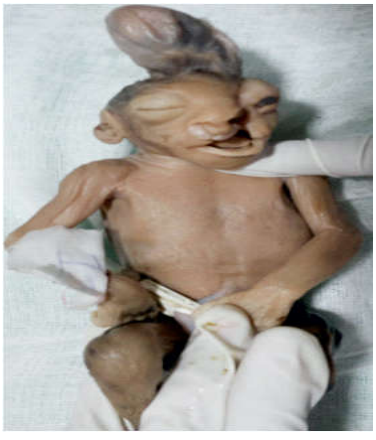

Fig. 2: Exencephalic Fetus with Cleft palate and cleft lip

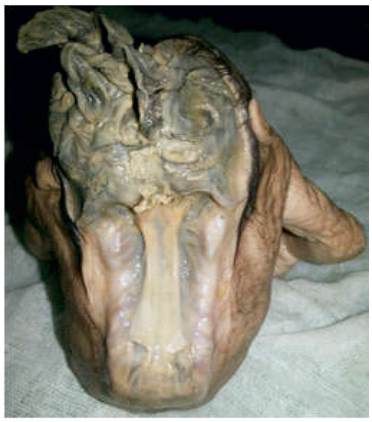

Fig. 3: Exencephalic Fetus with craniorachischisis

Fig. 4: ExencephalicFetus with craniorachischisis and kyphosis of thoracic spine

Fig. 5: Cardiovascular defects in ExencephalicFetus showing ASD

Fig. 6: Cardiovascular defects in ExencephalicFetus showing VSD

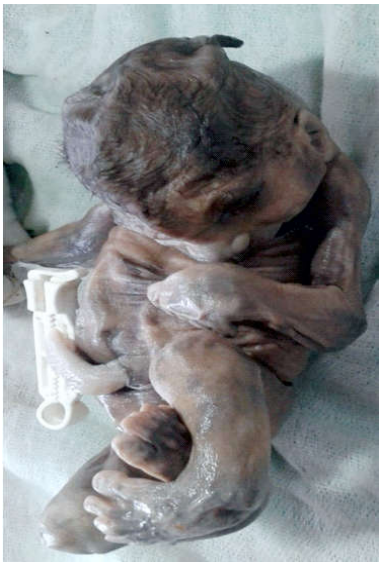

Fig. 7: Fetus with exencephaly showing clubbed feet and clubbed hand

Discussion

In exencephaly there is a large amount of recognizable neural tissue which is found outside the cranial vault. Facial views reveal frog-like appearance with prominent bulging eyeballs [12]. Chaurasia has given simpler classification of anencephaly based on occipito-vertebral and parieto-occipito-vertebraldefects[13]. Most of the studies suggests that if the defect is limited to the vault then it is classified as anencephaly and when the neural tissue protrudes through the defect it is called as exencephaly[3,4]. Exencephaly is also called as acrania .It has been proposed that the brain tissue of exencephalics may gradually degenerate due to the exposure to amniotic fluid in combination with mechanical trauma. This wearing down of the brain stroma produces the classic anencephalic features with flattened brain remnants behind the prominent orbits [14]. In the present study 11 fetuses out of 20 anancephalic fetuses were exencephalic and all the fetuses showed protruded eyeball, absent neck, folded ears and 6 fetuses showed protruded tongues (Figures 1, 2). Exencephaly can be detected on ultrasound scan at gestational age of 13 to14 weeks [5]. Exencephaly is often associated with rachischisis and other congenital defects [15]. Some animal studies have suggested that exencephaly/anancephaly was associated with malformations of the axial skeleton and spina bifida [16]. In the present study we found 4 cases of spina bifida with 3 case of craniorachischisis (Figures 3, 4) and 1 case of lumbar spina bifida. Also there is 1 case of kyphosis (Figure 3) and also 4 fetuses

showed club foot and clubbed hand. Neural tube defects may also presents with large thymus, small adrenal glands, hypo -plastic lungs, cyclopia, syndactyly, absent radius and thumbs, club foot, imperforate anus, cleft palate, renal and cardiac anomalies. It can also be associated with meningomyelocele, hydrocephalus and Chiari II malformation [17]. In the present study, gastrochisis, umbilical hernia was seen in 1 case each and in another case it was associated with omphalocele. There were 4 cases of cleft lip and cleft palate fig. Major defects of the cardiovascular system occur in 4 to 15 percent of live-born infants with anencephaly [2]. In the present study cardiac abnormalities were seen in 5 cases which were found to be the cases of ASD (3) and VSD (2) (Figures 5, 6).

Conclusion

Neural tube defects may be associated with the unbalanced form of a structural chromosomal abnormality in some families [18,19].Vitamin A deficiency, chromosomal trisomy (Ts12) [20], folate deficiency [21] can induce exencephaly. This study shows that associated morphologic anomalies are very common in fetus with neural tube defects. The study has described in detail the severe form of anencephaly that is exencephaly and the associated malformations. Therefore, considering the increase in incidence of the neural tube defects and the fatal abnormalities associated with it the knowledge of such may be important for academic, surgical as well as radiological procedures and for the accurate diagnosis of the condition. The present study highlights the same and will aid in the early and accurate diagnosis of the condition . The need of the hour is to take preventive measures to reduce the cases of NTDs. If screening and induced abortion are uniformly applied, the incidence of live-born infants with anencephaly would be less [2]. This can be done by interventions such as educational programmes and folic acid supplementation during reproductive age.

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