DOI: 10.7860/JCDR/2014/10842.5326

Case Report

Anatomy Section

Occipital meningoencephalocele with Cleft Lip, Cleft Palate and Limb Abnormalities- A Case Report

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ABSTRACT

A 21-week-old still born female fetus with occipital encepholocele, cleft lip and cleft palate was received from the Department of Obstetrics and Gynecology, Mahatma Gandhi Medical College and Research Institute, Pondicherry and was studied in detail. It was born to Primigravida, of a second degree consanguineous marriage, with unremarkable family history. The biometric measurements were noted which corresponded to the age of the fetus. Further the fetus was embalmed and dissected. On examination an encephalocele of 2.7×1.5 cm was seen in the occipital region with a midline defect in the occipital bone and herniated brain tissue. Other anomalies observed were right unilateral cleft lip, right cleft palate, and bilateral syndactyly of the lower limbs and associated Congenital Talipus Equino Varus of the right foot. Other internal organs were developed appropriate for the age of the fetus.

Keywords: Congenital talipes equino varus, Cleft lip, Cleft palate, Occipital meningoencephalocele, Syndactyly, Ultrasonogram

CASE REPORT

A 21-week-old still born female fetus with occipital encepholocele, right cleft lip and cleft palate was received in the Department of Anatomy, Mahatma Gandhi Medical College and Research Institute, Pondicherry and the same was studied in detail. It was delivered to a Primigravida, of second degree consanguineous marriage, with Non specific family history. The mother was 20-year-old. Folic acid tablet was taken as per routine during pregnancy. No history of fever with rashes, excessive vomiting, drug intake or radiation exposure. Second trimester scan showed the fetus in breech presentation. Calvarial outline was irregular. A mass was seen adjoining the cephalus [Table/Fig-1]. Liquor was adequate. Patient was advised termination of pregnancy following which fetus was expelled at 12 pm on 25/5/12 with consent of the parents. The biometric measurements were noted [Table/Fig-2] and it corresponded to the age of the fetus. An X ray of the fetus was taken [Table/Fig- 3]. No gross bony abnormalities of limb bones and ribs were observed. Skull bones could not be made out clearly. The fetus was embalmed following which gross examination was carried out and later dissected.

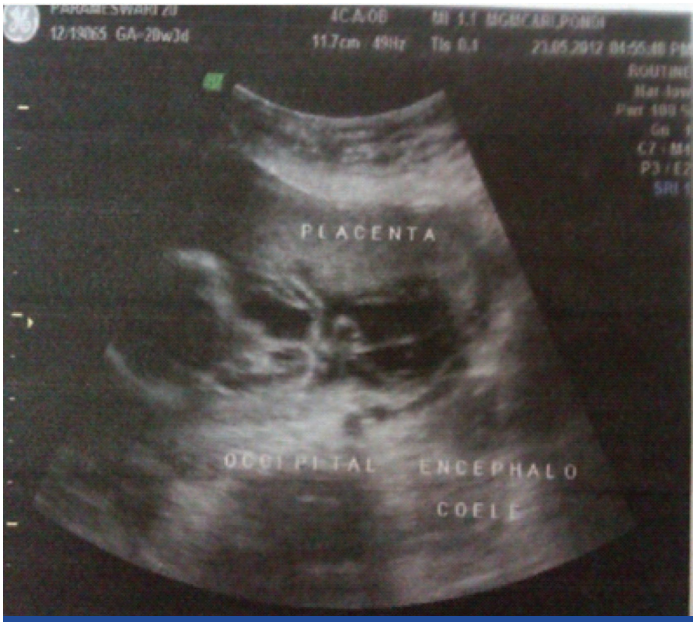
Detailed examination of the fetus showed the following abnormalities. A cystic mass measuring 2.7×1.5 cm was seen in the occipital region with a midline defect in the occipital bone [Table/Fig-4]. The Squamous part of the occipital bone was not fused in the midline above the foramen magnum resulting in the defect [Table/Fig-5]. Part of the tissue in the region of pons and cerebellum was seen to have herniated into the encephlocele sac. The vertebral column and the spinal cord were well developed [Table/Fig-6].

Other anomalies observed were right unilateral cleft lip and cleft palate [Table/Fig-7]. The right foot showed syndactyly of first and second toes and the left foot showed syndactyly of first, second and third toes [Table/Fig-8]. Base of the great toe of both the limbs showed a fibrous band attached to it [Table/Fig-8]. There was associated CTEV of the right foot [Table/Fig-8]. Other internal organs were developed appropriate for the age of the fetus.

DISCUSSION

Encephalocele is a congenital malformation characterized by a protrusion of the brain tissue and/or meninges through a skull defect [1]. Reportedly occurs in 0.8–5.6 per 10,000 live births [1,2]. The origin of the encephalocele is considered to be complex, and any associated risk factors have not been clearly identified [1]. The primary abnormality in the development of an encephalocele is a mesodermal defect resulting in a defect in the calvarium and dura associated with herniation of CSF, brain tissues and meninges through defect. Commonest site of encephalocele is occipital (75%), followed by frontoethmoidal (13% to 15%), sphenoidal or parietal (10% to 12%) [3]. Genetic factors, maternal nutritional deficiencies, and other environmental factors may facilitate the development of an encephalocele [1,4,5].

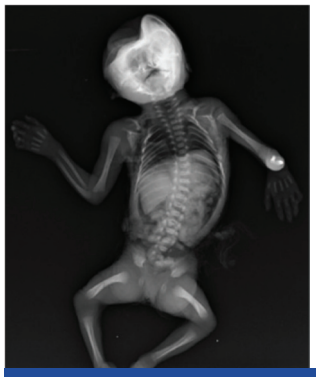
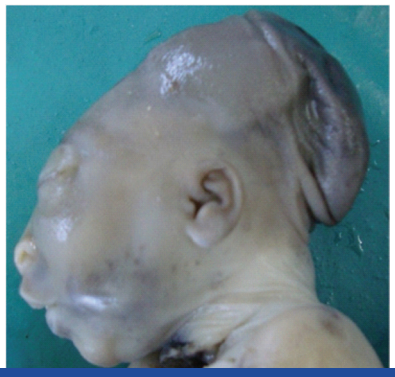
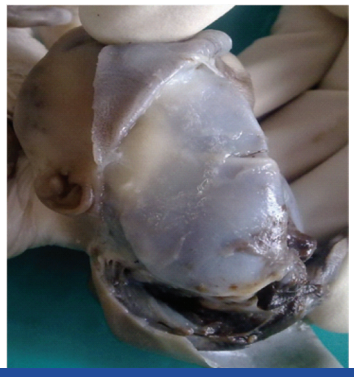
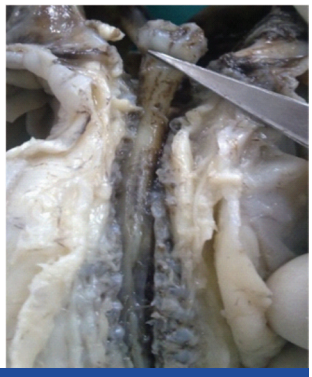
Occipital encephalocele presents as a mass in the occipital region usually covered by skin. They are often associated with other midline



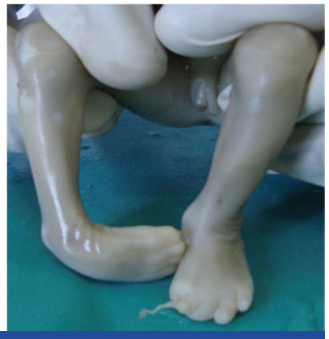
[Table/Fig-1]: USG showing occipital encephalocele

Biometric Parameters	Measurement (in cms)
Head circumference	15
Chest Circumference	13.2
Abdominal Circumference	11.8
Crown Rump Length	23

[Table/Fig-2]: Table showing biometric measurements of the fetus

[Table/Fig-3]: X ray of the fetus showing no gross anomalies of limb bones and ribs **[Table/Fig-4]:** Showing occipital encephalocele **[Table/Fig-5]:** Showing defect in occipital bone **[Table/Fig-6]:** Well developed spinal cord

[Table/Fig-7]: Cleft lip and palate **[Table/Fig-8]:** CTEV of right foot, syndactyly and fibrous band on the base of left great toe (arrow)

anomalies such as hypertelorism, broad nasal root, cleft lip, and cleft palate [6]. Other associations include microcephaly, microphthalmia, cleft lip and palate, polydactyly, polycystic kidneys and ambiguous genitalia. These features are typically seen in recessively inherited disorder Meckels syndrome [7].

Occipital encephaloceles occur due to a defect in fusion of occipital bone. The occipital bone develops from two sources. The paracordal cartilage surrounding the cephalic part of the notochord fuses to form the basal plate. It is then continuous with the occipital sclerotomes, the laminae of which meet behind forming the foramen magnum and squamous part of the occipital bone. Interparietal part of the occipital bone develops from membranous ossification [8]. The failure of fusion of these two parts of occipital bone has resulted in the defect in this case. Such midline defects are associated with other midline lesions [9]. This fetus presented with right cleft lip and cleft palate. Limb defects like radial, tibial bone aplasias and polydactyly associated with encephaloceles have been reported in literature [10]. This case presented with CTEV of right foot, syndactyly of toes, and presence of fibrous bands on both great toes. Such variable association of midline defects and limb defects with occipital encephalocele can be attributed to multifactorial aetiology.

The presence of fibrous bands on both great toes can be due to incomplete resorption of Apical Ectodermal Ridge (AER). Environmental and genetic factors have been implemented as a cause [11,12]. More than 80% of encephalocele cases are not associated with a certain genetic or chromosomal abnormalities [1]. Female predominance has been reported in literature [1,13,14].

80 to 90% of enchepaloceles in the Western hemisphere are occipital, anterior localization occurs much more commonly in the Eastern hemisphere [3]. Mahapatra et al., [15] and Hoving et al., [16] have reported higher incidence of anterior encephaloceles in South East Asia.

The prognosis of patients born with occipital encephalocele depends on the size of the defect and the amount of brain tissue herniated into the encephalocele. Surgical exicision of the sac followed by repair of the dural and the cranial defect is the treatment of choice [17]. But with associated anomalies the patients' prognosis becomes rather poor. Hence there is a need for early prenatal diagnosis of such congenital defects. Parents having a family history of occipital encephalocele with additional risk of consanguineous marriage should be monitored carefully [12]. The choice of termination of pregnancy depends on the amount of brain tissue herniated into the sac and associated anomalies. Encephaloceles with minimal brain tissue herniation have excellent prognosis [3,14]. MRI scan is recommended in such cases.

Hence during counseling sessions to couples with such risk factors the above mentioned prognostic indicators should be explained in detail. Limited literature regarding the aetiology and risk factors associated with occipital encephaloceles warrants additional prospective studies with larger populations. There is a need for development of a good surveillance program with a full proof reporting system. In fact in the surveillance manual for congenital anomalies developed by WHO, ICBDSR and CDC encephalocele, cleft lip and cleft palate have been included not only because of their ease of diagnosis but also because there is a potential for prevention, early diagnosis and treatment [18].

CONCLUSION

Association of occipital encephalocele with other anomalies was much different in this case from that reported in literature which can be attributed to the various environmental and genetic factors.

As encephaloceles are associated with high mortality and morbidity, their aetiology should be identified in detail for reducing worldwide incidences. Meticulous antenatal scan followed by careful history taking and clinical examination of the parents is warranted in these cases. Family history plays an important role in prognosis of encephaloceles. This will help in deciding the outcome of pregnancy and also facilitate parent counseling.

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FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **Aug 12, 2014**
Date of Peer Review: **Sep 16, 2014**
Date of Acceptance: **Oct 25, 2014**
Date of Publishing: **Dec 05, 2014**