

## Case Report

# Craniocerebral and Spinal Dysraphism with Omphalocele – A Case Report of Primary Neurulation Defect

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## ABSTRACT

**Background:** Anencephaly and Spina bifida are the two most common types of neural tube defects (NTDs). Disrupted formation and closure of neural folds leads to Craniocerebral and spinal dysraphisms.

**Materials and methods:** An 18 week old foetus was received in the department of Anatomy after elective medical termination of pregnancy due to the diagnosis of neural tube defect and associated congenital anomalies.

**Case Report:** The foetus had anencephaly, thoracic-lumbar spina bifida, omphalocele and clubbed foot and hands. The foetus was dissected and studied, to look for abnormal internal structures. On meticulous dissection it was found that there were abnormalities in spine, gastrointestinal system and cranium.

**Conclusion:** Anencephaly is a neural tube defect which has multiple neural and non-neural associated anomalies. A detailed description of the combination of associated anomalies goes a long way in updating knowledge on the same.

**KEY WORDS:** Anencephaly, spina bifida, omphalocele, folic acid, neural tube defects.

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## INTRODUCTION

The central nervous system develops from a streak of ectodermal cells which are present on the dorsal aspect of the embryo that specialize into neuroepithelium. It starts as a

plate of cells that folds into the neural tube. Disrupted or arrested development of these neural structures, also called dysraphism, are connected with the disturbances in formation of overlying mesodermal (muscle, bone, dura)

and nonneural ectoderm (cutaneous) structures also. The spectrum of anomalies due to cranio-cerebral dysraphism include, anencephaly, craniorachischisis, encephaloceles. Those due to spinal dysraphism include myelomeningocele, myeloschisis [1]. Craniorachischisis is a condition marked by a neural tube defect that results in the absence of the majority of the skull and brain (anencephaly) combined with an open vertebral column (spina bifida). It is also defined as a caudal extension of anencephaly [1].

The embryonic exencephalic brain undergoes abnormal vascularisation due to the failure of closure of neural tube during the 4<sup>th</sup> week of development [2]. Due to this the developing brain undergoes degeneration and remains as a spongy mass which is vascular with degenerated hind brain structures leading to neural tube defects [3]. Anencephaly and Spina bifida are the 2 most common types of neural tube defects (NTDs), are estimated to affect around 3000 pregnancies each year in the United States [4]. Race and ethnicity also influence the prevalence of NTDs, highest rates seen in Hispanic ethnicity and least in the Asians [5]. With nutritional supplementation and folic acid fortification the incidence of Dysraphism has markedly reduced by 24% in a span of 7 years between 1994 and 2001 in the United States of America [6].

### METHODS

An 18 week old foetus with developmental anomalies was received in the department of Anatomy, Sree Balaji Medical College And Hospital, Chennai, India. It was fixed in formalin solution after the medical termination. The external features were observed. Dissection was done to study the internal features and abnormalities were noted. Retrospectively the history of the intrauterine life of the foetus was traced and the mother's antenatal history was retrieved from the records. The findings in external features and internal features were noted and compared with previous such cases.

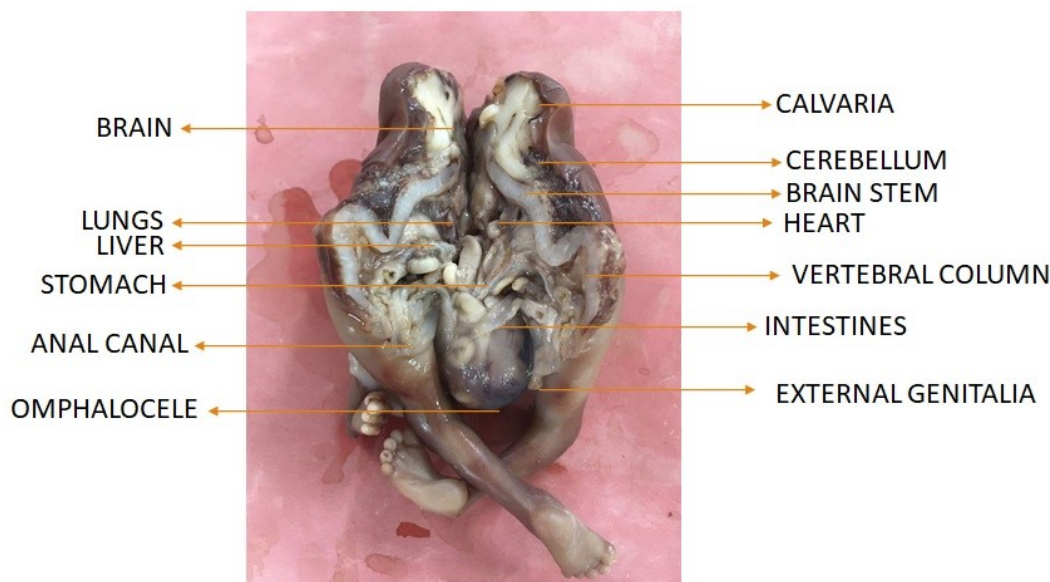
### CASE REPORT

The foetus received in the Department of Anatomy was about 18 weeks gestational age.

The mother of the foetus was a 24 year old lady, this was her first gravida. She sought for obstetric consultation at 15 weeks, she was not on nutritional supplementation or fortification. On routine obstetric ultrasound scan she was found to have a foetus with neural tube defect and was counselled medical termination. The patient revisited at 18 weeks and gave consent for termination after which a medical termination of pregnancy was done at 18 weeks.



**Fig. 1:** External features of an anencephalic foetus. 1a: facial features, 1b: low set ears, omphalocele, clubbed foot and hands, 1c: thoraco-lumbar spina bifida



**Fig. 2:** Dissected with a posterior mid-sagittal incision. Shows S shaped bend in the vertebra, absence of bony calvaria herniated gut in the omphalocele, anteriorly dislocated heart and lungs.

On detail examination of external features (fig 1), it was found that, the total length of the foetus was 11cm, the crown-rump length: 5.7cm, which corresponded to only 12+3 weeks gestational age. Head circumference is: 7cm. There was absence of bony calvaria, cranium was covered by skin only. Low set ears were present close to the shoulder on both the sides. Neck was absent. Mandible was maldeveloped. There was a Lumbo-Sacral Spina Bifida with meningomyelocele. Length of spina bifida: 2.8cm and width: 0.8cm. There was presence of omphalocele in the anterior abdominal wall with protrusion of abdominal contents through the umbilicus. Due to the presence of omphalocele the anterior abdominal wall was not fused completely in the midline along the linea alba. The abdominal wall defect measures 2.2 cm in width and 3.5 cm in length. The omphalocele contained the loops of jejunum and ileum. Umbilical cord is observed emerging from the left side of the omphalocele, with umbilical blood vessels. External genitalia was that of a male with normally developed penis, empty scrotum and undescended testis. The femur length was 2.8cm. The foetus had clubbed foot on the left and clubbed hand on the right.

On dissection (fig: 2), the cerebrum and cerebellum are poorly developed. The spinal cord is not well differentiated beyond the cervical level. There is an S-shaped curve in the vertebra at the Thoraco-Lumbar region. At

the thoracolumbar junction the vertebra was deeply notched anteriorly. The loops of ileum and jejunum along with the peritoneum has herniated through the abdominal wall defect. The kidney and ureter were normally placed. Scrotum was empty. With no genital or gonadal abnormalities. Organs like lungs and heart are anteriorly dislocated to the abnormal curvature of the vertebra. Liver spleen and stomach were appreciated, in their normal location and development. There was no associated cleft lip, cleft palate or facial clefts.

## DISCUSSION

Anencephaly is associated with many congenital anomalies, the range varying from 9.4% in some to 84% in others [7,8]. The incidence of anencephaly is 1 to 9 / 1,000 births, and found to be highest in the U.K and Ireland [9].

The common associated anomalies include spina bifida, cleft palate, cleft lip, clubbed foot and clubbed hands, genital anomalies, gut anomalies. In our case report the foetus had spina bifida, omphalocele and clubbed left foot and right hand. In one study on anencephaly and associated malformations by Ashok Gole et al, spina bifida is seen in 45% of anencephaly fetuses. Omphalocele in 5% and clubbed foot and hands in 35% [10].

There has also been studies suggesting female preponderance in craniocerebral and spinal dysraphisms [8,10-12].

Risk factors for anencephaly are multiple. Anencephaly is considered to have polygenic inheritance with environmental factors influencing it additionally. The interaction of genes that predisposes to neural tube defects are yet to be identified [9]. Folic acid is considered to have an external effect on Neural tube defect, because folic acid supplementation has found to decrease its incidence. This can either be due to dietary causes or impaired folic acid metabolism [4,9,13,14]. In this case the mother came for obstetric consultation only at 15 weeks and hence was not on early folic acid supplementation. Maternal obesity is also considered a risk factor for anencephaly and spina bifida. Pre pregnancy body mass index greater than 29, is a chief contributory risk factor [15,16].

### CONCLUSION

Dysraphisms are a group of congenital anomalies which present with various associated anomalies, a detailed description of the combination of associated anomalies goes a long way in updating knowledge on the same. A detail dissection and microscopic studies on the organs of such fetuses terminated due to neural tube defects can be done to look for the scope of organ harvesting for transplantation. Hence reporting cranio-cerebral dysraphisms is important in multiple fields of medicine.

**Conflicts of Interests:** The authors declare that there are no competing interests

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