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# Neural Tube Closure Failure: A Case Series

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***Abstract:***
***Background:*** *NTDs arises due to incomplete closure of neural tube during early embryonic development. The abnormal development leads to miscarriage, stillbirth and abnormalities of CNS in individuals who survived. NTDs are classified according to types and position of the lesion.*
***Methodology:*** *Fetuses with NTDs were collected from Dept of OBG RIMS for a period of 1 year after taking permission from the concern authority and parents.The sex, gestational age, type of NTDs and associated anomalies were observed and documented.The required data were collected and documented.*
***Results:*** *The incidence of NTD was found to be 0.1%. Out of 10 NTDs, the most common was anencephaly followed by spina bifida. On examination: Fetus 1 shows anencephaly with protruding eye, cleft lip and cleft palate and placenta cephalica. Fetus 2 and Fetus 3 shows encephalocele at different sites. Fetus 4 shows buldging over sacrococcygeal region, vertebral dysmorphism showing spina bifida. Fetus 5 shows meningocele along with Arnold chiari malformation type II. Fetus 6 shows absence of upper part of cranium with herniated brain indicating exencephaly.*
***Conclusion:*** *NTD is the most common congenital malformation that often lead to spontaneous abortion. The present study found the morphological diversities of the NTD, the commonest of which is Anencephaly. Early detection by prenatal USG and prevention by folic acid prophylaxis is important because of its high incidence.*
***Keywords:*** *Neural tube defects (NTDs), Anencephaly, Spina bifida, Prenatal diagnosis, Folic acid prophylaxis*.
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## I. Introduction

Neural tube defects (NTDs) are severe congenital malformations of spinal cord that result from the failure of closure of the neural tube during the 3rd to 4th week of embryonic development. The resulting abnormalities may involve the meninges, vertebrae, muscles and skin.[1,2]The NTDs are broadly classified into cranial and spinal defects. If closure fails in the cranial region, it results in anencephaly, hydranencephaly, encephalocele, iniencephaly. The anencephaly is further subdivided into microencephaly and holoencephaly. The spinal defects are classified into open and closed neural defects. Open variety includes myelocele and meningomyelocele and closed variety includes meningocele and spina bifida occulta.[3] Open NTDs are the most severe and occur in about 0.1% of all live births.[3]NTDs are associated with many malformations. The most common associated malformations are other central nervous system or spinal defects, spinal deformities, club foot and cleft lip or palate.[5]These malformations expose neural tissue to the external environment leading to neurological impairment.[6]NTDs have no single genetic or teratogenic cause and are believed to be multifactorial, that is from the interaction of both genetic and environmental factors and the frequency varies by race, which suggest genetic predisposition.[3]The Global prevalence of NTDs is estimated to be 0.5 – 2 per 1000 pregnancies[7], however, the prevalence of NTDs in India is very high, approximately 4.5 per 1000 live births.[2]Worldwide approximately 300,000 babies are affected annually resulting in 88,000 deaths & 8.6 million DALY.[4]Folic acid deficiency has been strongly implicated and periconceptional supplementation reduces the risk by upto 70%, although not all cases are preventable.[8]Despite advances in prenatal diagnosis & preventive strategies, NTDs continue to contribute significantly to perinatal mortality & long term morbidity.

## II. Material And Methods

The study was conducted at Department of Anatomy, Regional Institute of Medical Sciences, Imphal for a period of 1 year May 2024 to May 2025. Fetuses with NTDs were taken from terminated pregnancies and

stillbirths from the Department of Obstetrics and Gynaecology. Convenience sampling method was done. Fetuses were observed in detail morphologically for the sex, type of NTDs and other associated anomalies and the findings were documented. Number of total deliveries (terminated pregnancies, stillbirths and live births) and number of fetuses with NTDs were also documented.
Research ethics board committee permission was obtained.

## III. Result

The median gestational age of fetuses with NTDs was 19 weeks ranging from (13 – 26) weeks. Number of deliveries during the study period was 8506. Out of the total deliveries, the no. of fetuses with NTDs were found to be ten. The incidence of NTDs in the present study was found to be 1.17 per 1000 births. The most common NTD was documented to be anencephaly (40%) followed by spina bifida (30%). Out of 10 NTDs, associated malformations were observed in 3 (30%) cases.Table 1. shows that the most common NTD is anencephaly (40%) followed by spina bifida (30%) Table 2. shows that the associated malformations in the present study were clept lip, clept palate, placenta cephalica, Arnold chiari malformations. Amongst the associated malformations, cleft lip and cleft palate were commonest.Fig1. shows a fetus of 14 week gestation with anencephaly associated with protruded eyeball, cleft lip and cleft palate along with placenta cephalica. Fig 2. shows a fetus of 13 week gestation wih external bulging over the sagittal fronto-parietal region suggesting anterior encephalocele. Fig 3. shows a fetus of 16 week gestation with external bulging on the mid parieto-occipital region suggesting posterior encephalocele. Fig 4. shows a fetus of 22 week gestation with external bulging (3X3cm) overlying lumbosacral region which on MRI showed vertebral dysraphism (kyphosis), on dissection, the mass contained meninges and spinal cord suggestive of meningomyelocele.Fig 5. shows fetus of 24 week gestation which has a bulging over sacrococcygeal region (3x3 cm), on dissection, the bulging contained meninges and neural tissue and the MRI showed downward displacement of cerebellum and medulla oblongata suggestive of myelomeningocele with Arnold- Chiari Malformation type II.

Fig 6. shows fetus of 26 week gestation with absence of upper part of cranium along with herniated brain indicating exencephaly associated with cleft lip and cleft palate.

**Table 1: Distribution of the types of Neural Tube Defect**

| Type of NTD | | No. of fetuses (N= 10) |
|---|---|---|
| Cranial | Anencephaly | 4 |
| | Exencephaly | 1 |
| | Encephalocele | 2 |
| Spina bifida | Myelomeningocele | 2 |
| | Spina bifida occulta | 1 |

**Table 2: Neural tube defects with gestational age and associated malformations.**

| Types of NTDs | Gestational age | Associated anomalies |
|---|---|---|
| Anencephaly | 14 weeks | Cleft lip, cleft palate and placenta cephalica |
| Meningocele | 20 weeks | Arnold chiari malformation type II |
| Exencephaly | 26 weeks | Cleft lip, cleft palate |

Fig1: Anencephaly with protruded eyeball, cleft lip and cleft palate and placenta cephalica

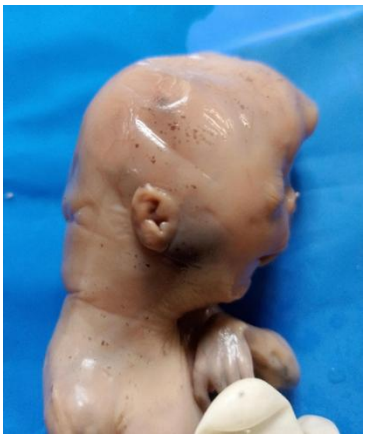

Fig 2: Fetus with anterior encephalocele.

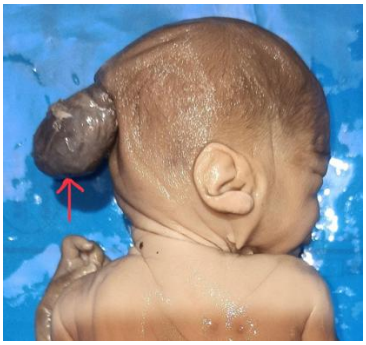

Fig 3: Fetus with posterior encephaloce

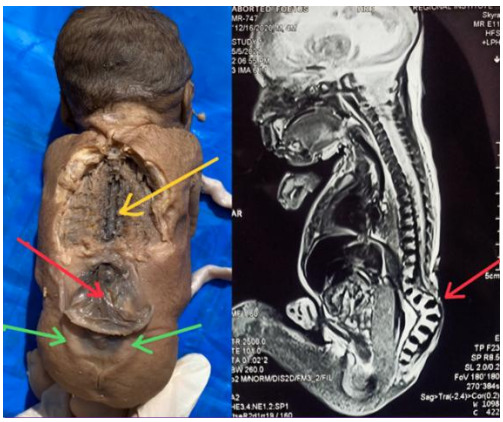

Fig 4: Fetus with meningomyelocele.

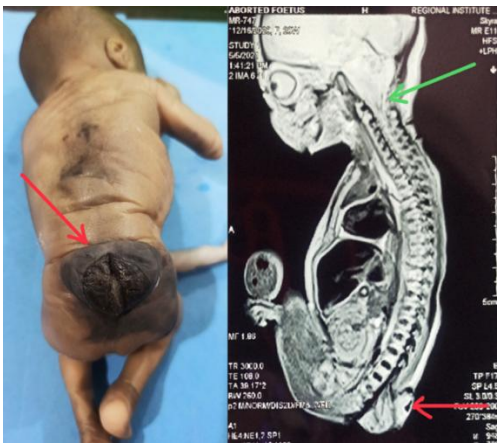

Fig 5: Fetus with myelomeningocele and Arnold- Chiari Malformation type II

Fig 6: Exencephaly with cleft lip and cleft palate

## IV. Discussion

Neural tube defects(NTDs) are among the most common congenital malformations of the central nervous system resulting from failure of neural tube closure during early embryogenesis, typically between the 3rd and 4th week of gestation. These defects contribute significantly to fetal morbidity, mortality and spontaneous abortions.

In the present study, the incidence of NTDs was 1.17 per 1000 births, which falls within reported range for India (0.5-11 per 1000 births) and is comparable with the global prevalence of approximately 1-2 per 1000 births, similarly in a study by Menasinkai SB[11] reported an incidence of approximately 1.06% in an Indian population, further supporting the variability in prevalence across different populations.

Among the different types of NTDs observed in this study, anencephaly was the most common(40%), followed by spina bifida(30%) and encephalocele(30%). A similar observation was reported by Mailaram S et al[4], who also found anencephaly to be the most prevalent anomaly among NTDs. However, Moradi B et al[5] reported spina bifida as the most common anomaly, followed by cephalocele and anencephaly. Likewise, Ssentongo P et al[9] reported a higher birth prevalence of spina bifida compared to anencephaly and encephalocele in their meta-analysis. These differences in the pattern of NTD distribution may be attributed to variations in geographical location, nutritional status, genetic predisposition and environmental exposures.

The median gestational age of the fetuses in the present study was 19 weeks, ranging from 13 to 26 weeks, which corresponds to the period when most severe congenital anomalies are detected through routine antenatal screening. Moradi B et al[5] also emphasized the importance of prenatal ultrasonography in the early detection and classification of NTDs, highlighting its role in improving pregnancy management.

In the present study, associated congenital anomalies were observed in 30% of cases, with cleft lip and cleft palate being the most common associated malformations. NTDs are frequently associated with other congenital anomalies due to disturbances occurring during early embryological development. A case report by Jamous MA et al[12] described the rare occurrence of multiple NTDs in a single patient, emphasizing the need for early surgical intervention to prevent complications.

Among cases of spina bifida, associated abnormalities such as Arnold-Chiari malformation and hydrocephalus have been reported in earlier literature. In the present study, Arnold-Chiari malformation type II was observed in one case (10%), which supports the established association between myelomeningocele and hindbrain herniation.

The etiology of NTDs is multifactorial, involving a complex interaction of genetic and environmental factors. Endalifer ML et al[10] reported that the epidemiology and determinants of NTDs vary widely across different countries, geographical regions and socioeconomic groups. Isakovic J et al[13] also highlighted the role of gene-environment interactions, including folate deficiency, exposure to teratogens and maternal hyperthermia in the development of NTDs.

Several studies have emphasized the importance of folic acid supplementation in the prevention of NTDs. Blencowe H et al[8], estimated that nearly 260,000 pregnancies are affected annually by NTDs worldwide, particularly in low and middle-income countries and concluded that adequate folic acid supplementation could prevent up to 70% of cases. Similarly, Heseker HB et al[13] reported that folate supplementation significantly reduces the risk of NTDs, while Czeizel AE et al[15] provided early evidence supporting periconceptional vitamin supplementation as an effective preventive strategy.

## V. Conclusion

Neural tube defects demonstrate a wide spectrum of severe congenital anomalies resulting from early neural tube closure failure. The present case series highlights the morphological diversity of NTDs, ranging from severe cranial defects such as anencephaly to spinal defects including various forms of spina bifida and associated

anomalies like cleft lip, cleft palate and Arnold-Chiari malformation. These findings emphasize the importance of neural tube closure in determining the type and severity of defects. Importantly, NTDs are largely preventable. Adequate periconceptional folic acid supplementation, early antenatal screening, and timely diagnosis through ultrasonography can significantly reduce their incidence and associated complications.

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