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Case Series Of Spinal Dysraphism In A Tertiary Care Centre

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Abstract

Neural tube defects (NTDs) are the second most common cause of congenital anomaly worldwide (cardiac anomalies is the first). The 3rd and 4th week of gestation is the critical period for neural tube development. Multiple genetic and environmental factors are known to cause the NTDs in a developing embryo. Survival of the newborn also depends on the severity of the lesion. Hydrocephalus is the commonest abnormality allied with NTDs in syndromic cases . Folic acid supplementation in all prospective mothers, preferably 4 weeks before conception and at least 12 weeks after conception, can prevent NTDs. Here we report 10 cases of neural tube defects being managed successfully in Tripura medical college & Dr. BRAM Teaching hospital.

Keywords: neural tube defects, hydrocephalus, folic acid

Introduction

Neural tube defects (NTDs) are anomalies of neurulation during fetal development. The NTDs are broadly classified into cranial and spinal defects. The cranial defects are further classified into encephalocele, anencephaly, and iniencephaly. The anencephaly is further subdivided into microencephaly and holoanencephaly. The spinal defects are classified into open and closed neural defects. Open variety includes myelocele and meningomyelocele. Closed variety includes meningocele and spina bifida occulta.[1] Most of these cases are diagnosed prenatally, and termination of pregnancy for fetal anomaly is the most common outcome for fetuses with NTD.[2,3] Recurrence risk after one affected pregnancy is 2%–3% for any open NTD. NTDs, serious birth defects of the brain and spine, are a major, preventable public health burden. Globally, it is estimated that approximately 300,000 babies are born each year with NTDs,[4] resulting in approximately 88,000 deaths and 8.6 million disability-adjusted life years.[5,6] Periconceptional folic acid supplementations have been shown to reduce the prevalence of open NTDs by at least 60%.[7] This reduction occurred both among mothers with previously affected pregnancies and among those who have no such risk factors. On the basis of many researches, it became apparent that women planning pregnancies should be advised to take a dose of 4 mg–5 mg folic acid for 2 months before conception and throughout the first trimester.[8] The present case series aims to report various types of spinal dysraphism, their mode of presentation and their management.

Materials And Methods

The study was conducted at the Department of general surgery, Pediatric surgery unit of TMC & DR.BRAM teaching hospital, Hapania, Tripura , for a period of 1 year from sept 2020 to sept 2021. All the newborn babies and also the infants with visible and occult spinal defects are included in the study, after confirming the NTD in patients and prior oral and written consent was taken. There were 7 male and 3 female patients. Out of which, 4 patients presented as a case of ruptured meningomyelocele and 5 patients presented with complain of a swelling at the lower

back since birth and 1 presented with sacral dimple. Out of the 10 patients, 8 patients had associated hydrocephalus , which responded to medical therapy, 1 of them needed VP shunting, 1 of them expired on the day of admission.

Institutional ethics committee permission was obtained for the present case series.

Cases

Case 1

A newborn baby of 4days old was referred to emergency of TMC & DR.BRAM teaching hospital, with the chief complaint of swelling at the lower back. The baby was admitted in the SNCU of TMC. Initial evaluation and stabilisation of the baby was done. Then this baby was referred to pediatric surgery unit of TMC. After further evaluation it was diagnosed to be a case of lumbo-sacral meningomyelocele with associated hydrocephalus. On further clinical evaluation , the lower limb motor function was found to be intact. The baby was prepared for NTD repair after thorough evaluation and ruling out other associated anomalies. Post - operatively , the baby was given acetazolamide to prevent any CSF leak and also to prevent hydrocephalus and the baby was evaluated daily by monitoring head circumference , tone of lower limbs, any episode of fever with chills and rigor, convulsions. The baby was on full breast feed by post-operative day 2 and was discharged successfully on POD -14 after completion of antibiotic therapy.

Fig 1- showing pre-operative lumbo-sacral meningomyelocele and post-operative repair of NTD.

Case 2

A 2days old neonate was referred to the pediatric surgery unit of TMC as a case of ruptured MMC. The baby was admitted in the SNCU of TMC. After initial resuscitation and evaluation , the baby was taken up for emergency NTD repair. Post-operative period was uneventful. The baby was discharged on post operative day 21 after completion of antibiotic therapy. On follow up, one month after the surgery, the baby presented with hydrocephalus which was non-responsive to medical therapy. At the age of 1 and half month this baby was again admitted for VP shunting. On post operative day 10 , the baby was discharged successfully and is doing fine till date.

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Fig 2-Pre-operative ruptured meningomyelocele with leaking csf and immediate post-op pics.

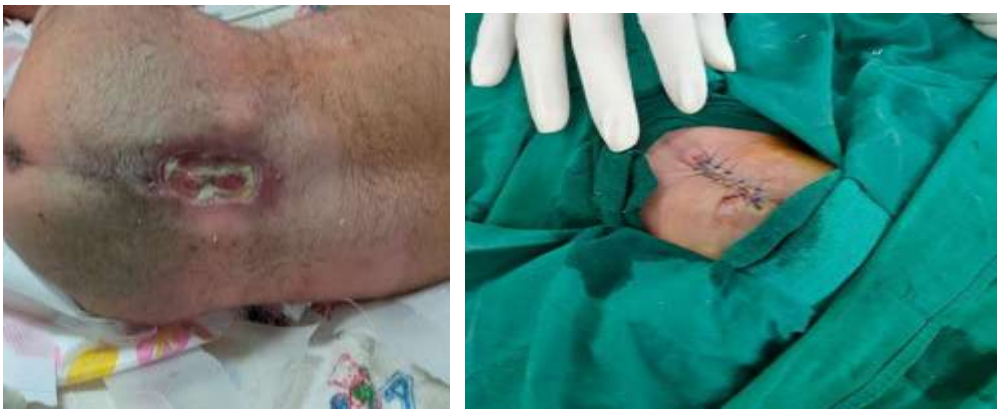
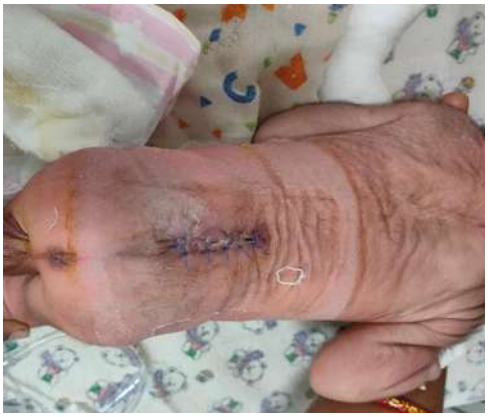


Fig 3 – post-operative day 5 of NTD repair.



Case 3

A 4 month old baby boy was referred from Makunda, as a case of huge lumbo-sacral meningomyelocele. The baby was admitted in pediatric ward under pediatric surgery unit. The patient was evaluated thoroughly and was planned for elective repair of neural tube defect. Post operative stay was uneventful and the patient was discharged after 2weeks of surgery. Post operative follow-up at 3weeks after the discharge shows no hydrocephalus with intact tone of lower limb muscles.

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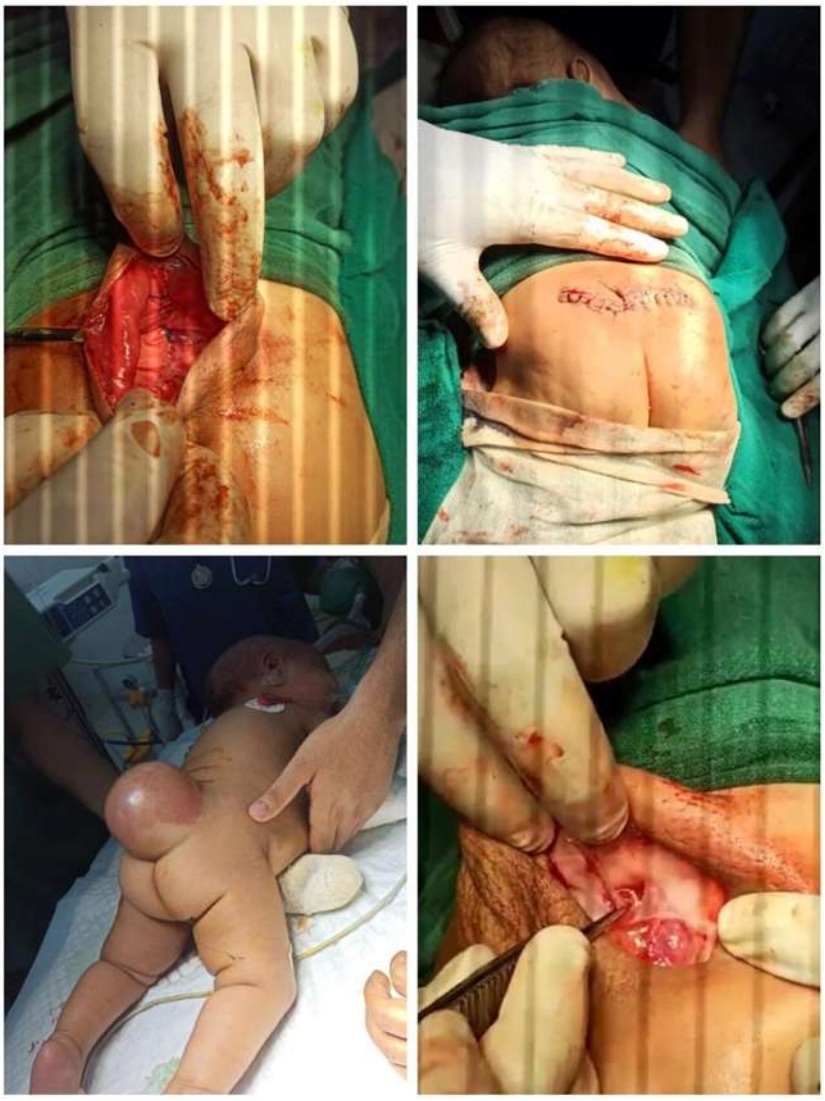
Fig 4- showing pre-op, intra-op, post-op pictures of huge lumbosacral meningomyelocele without any hydrocephalus.



Case 4

A 5days old baby was referred to the emergency of Tripura medical college and DR.BRAM teaching hospital as a case of ruptured meningomyelocele with leaking CSF. The baby was admitted in the SNCU of TMC and was referred to pediatric surgery unit. After initial evaluation and stabilisation , the baby was taken up for emergency NTD repair. The defect was repaired with prolene 5-0. Post operatively the baby develoed fever for which injectable antibiotics were upgraded to inj vancomycin and meropenem. The baby did well and was discharged on 3rd week after surgery.

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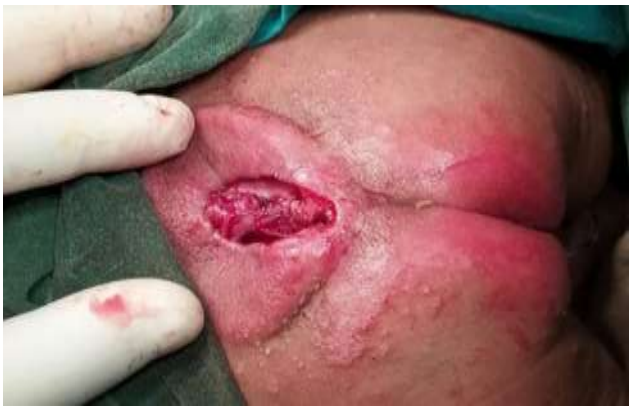
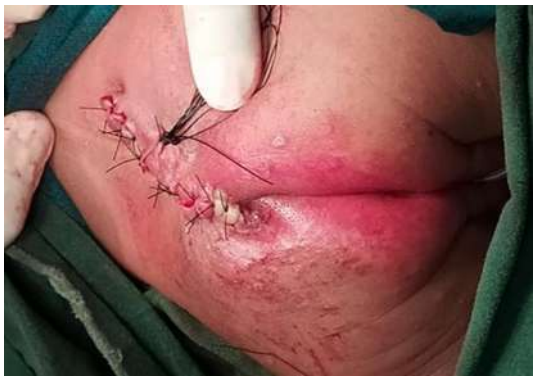
Fig- 5- showing ruptured lumbosacral meningomyelocele with leaking CSF.

Fig-6- showing intra-operative repair of neural tube defect and immediate post-operative skin closure.





Case 5

A 5days old baby with lumbo-sacral meningomyelocele with associated huge hydrocephalus and features of meningitis was referred to TMC and DR.BRAM teaching college emergency dept. The baby was admitted in SNCU , evaluated thoroughly and initial resuscitation was done with injectable antibiotics and intravenous fluids. The baby was toxic with high grade fever and had multiple episodes of seizure and convulsions. The baby expired in the evening of admission day even after all resuscitative measures.

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Fig 7- showing pic of the newborn baby with lumbosacral meningomyelocele with huge hydrocephalus.

Case 6

A baby of 6days old with lumbo-sacral meningomyelocele was referred to pediatric surgery unit of TMC and DR.BRAM teaching hospital. The baby was admitted in SNCU of TMC. After initial evaluation and stabilisation, the baby was taken up for operation. The operation lasted for 2hours. Intra-operatively, the spinal nerve roots herniating through the posterior arch of the vertebra was identified, safeguarded and was dissected meticulously. The neural placode was tubularised with prolene 5-0. The dura matter was then sutured in watertight manner so as to prevent any CSF leak. Lastly flap coverage of the defect was done with ethilon 3-0.

Post operative period was uneventful and the baby was discharged successfully on post operative day 14.

Fig-8- picture showing pre-operative lumbo-sacral meningomyelocele and intra operative dissection of neural placode.

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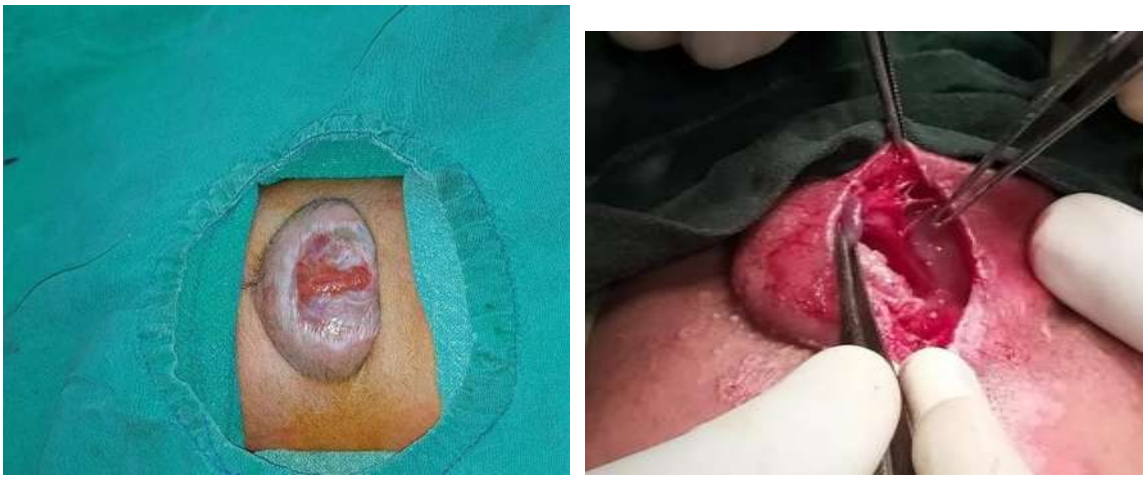
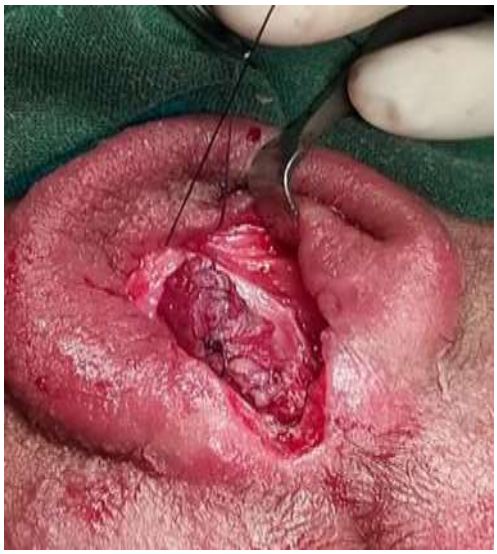


Fig-9- picture showing watertight closure of dura mater.



Case 7

A 10day old baby was referred to pediatric surgery unit of TMC and DR.BRAM teaching hospital as a neglected case of huge lumbo-sacral meningomyelocele with very poor lower limb tone. The baby was admitted in SNCU of TMC and was initially resuscitated with intravenous fluids and injectable antibiotics. After initial evaluation of the baby , the baby was prepared for operation. It took 3 hours to debride the infected wound and to fashion a flap for adequate coverage of the defect apart from closing the neural tube. Post operatively, the baby developed sepsis which was managed by upgrading the antibiotics to injectable meropenem and vancomycin. The baby was discharged successfully on post operative day 21.

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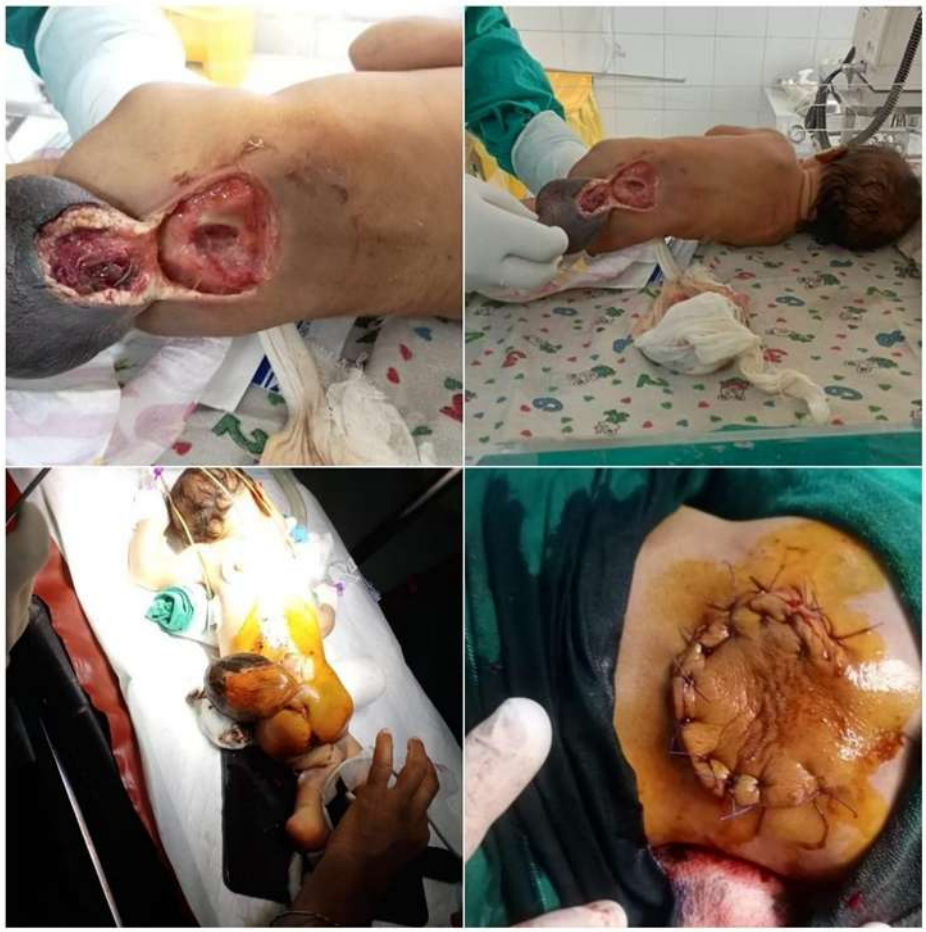
Fig-10- picture showing neglected case of lumbo-sacral meningomyelocele with gangrenous changes and its coverage with a transposition flap.



Case 8

A 3days old baby referred to SNCU of TMC as a case of lumbo-sacral meningomyelocele. The baby was admitted and was referred to pediatric surgery unit of TMC. The baby was evaluated and prepared for operation. The operation was uneventful and the baby recovered well. Post operatively the baby was discharged on day 14.

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Fig- 11-picture showing lumbo-sacral meningomyelocele and dissection of neural placode.

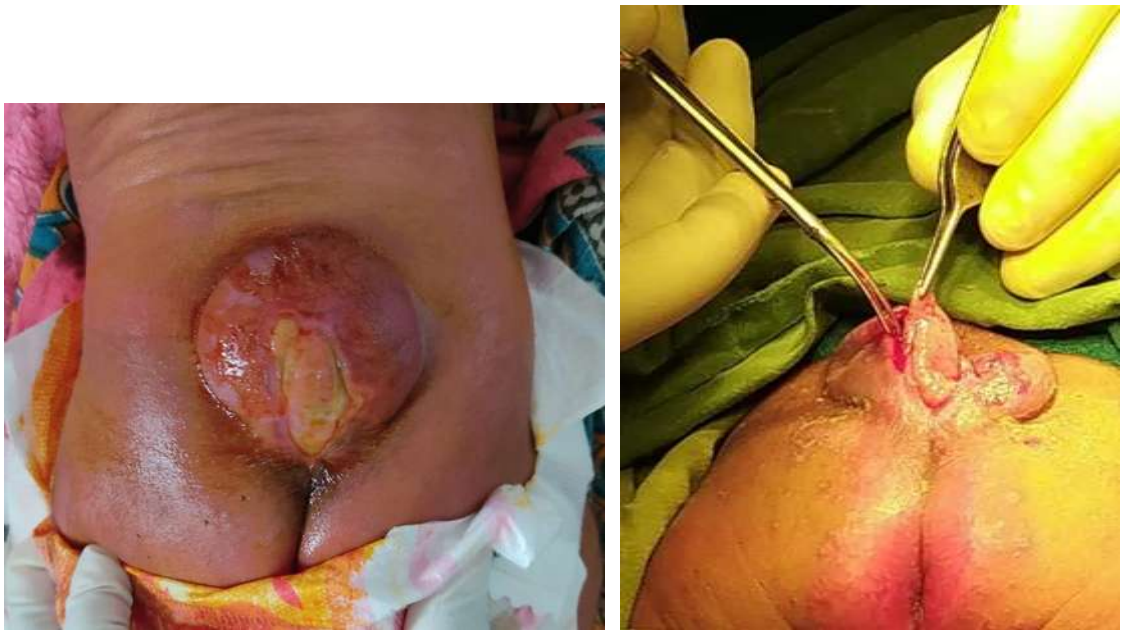

Fig -12 – picture showing closure of neural placode with prolene 5-0.

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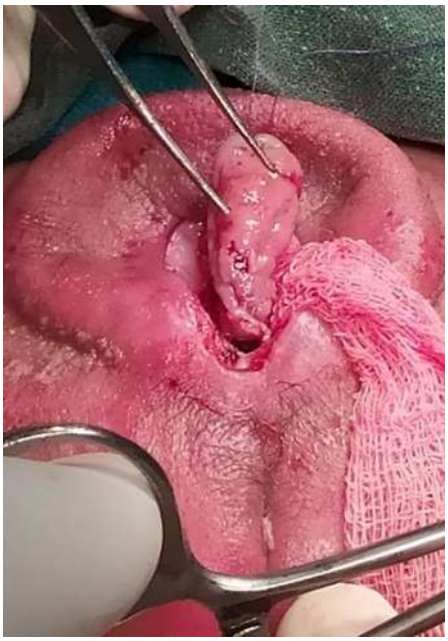

Case-9

A 3day old baby was referred from Makunda , Assam to the casualty of TMC and DR.BRAM teaching hospital as a case of swelling at the back since birth. The baby was admitted in SNCU of TMC and was stabilised by IV fluids and injectable antibiotics. After initial resuscitation and stabilisation of the baby, thorough evaluation was done and was diagnosed as a case of lumbo-sacral meningomyelocele. The baby was referred to pediatric surgery unit of TMC. After proper assessment of the baby , it was finally diagnosed as a case of lumbo- sacral meningomyelocele with communicating hydrocephalus with poor lower limb tone. The baby was planned for neural tube defect closure as soon as the investigations were available. Intra operatively , neural placode was repaired with prolene 5-0, without injuring any spinal nerves and also avoiding tension during the repair. The covering duramater is repaired with prolene 5-0. The skin gap is covered by surrounding flap (transposition flap).

Post operatively the patient was put on oral acetazolamide to prevent CSF leak and to prevent hydrocephalus. The baby recovered well but the tone of lower limb muscles remained the same as it was pre-operatively. The baby was discharged successfully after 2weeks of injectable antibiotics, on full oral feed.

Fig-13 – pictures showing preoperative meningomyelocele and intra operative repair of neural placode.

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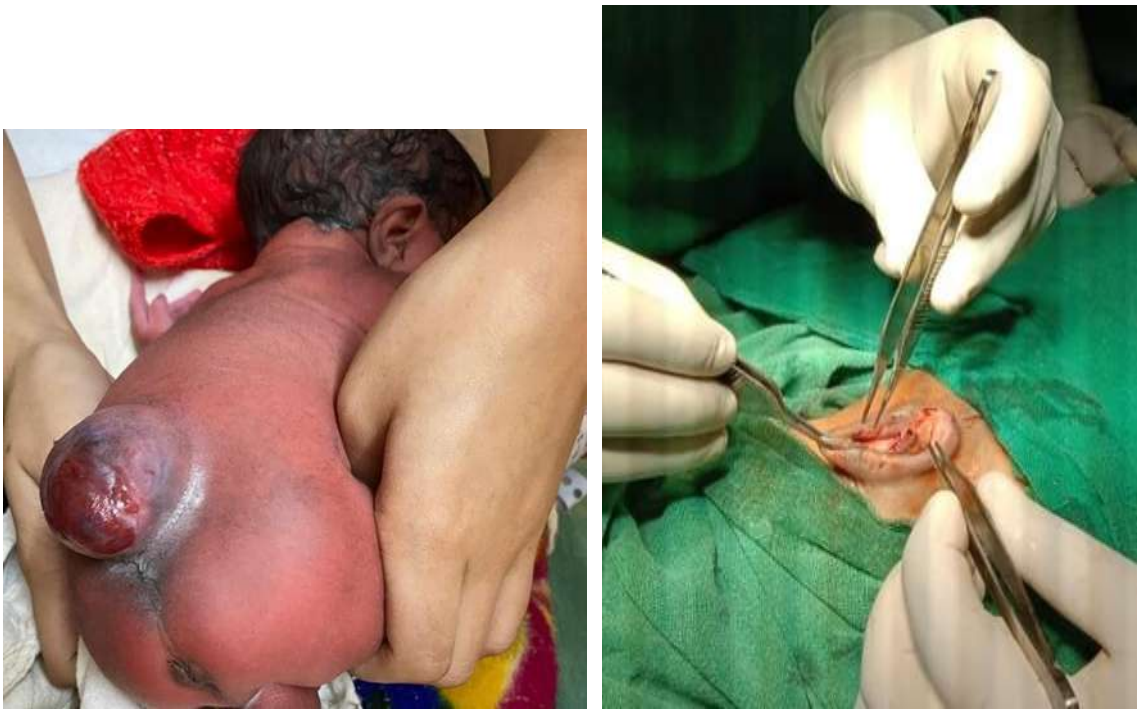
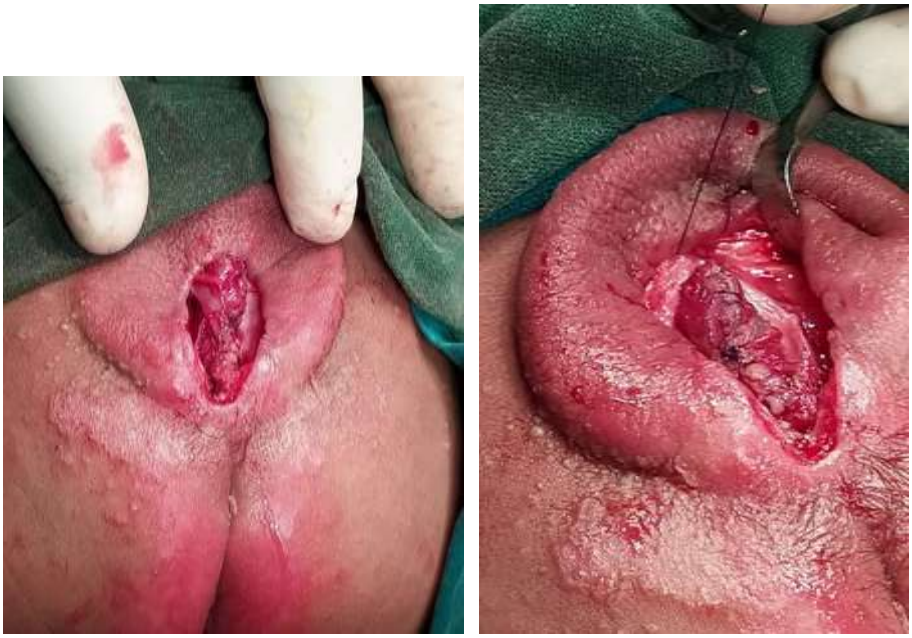


Fig-14- intra operative pictures showing dural cover of the neural placode.



Case-10

A baby of 2months old was referred to pediatric surgery unit of TMC and DR.BRAM teaching hospital with chief complaint of cleft in sacral region. The baby was clinically assessed and was admitted in PICU of TMC . MRI of lumbosacral region along with brain was done which revealed tethered cord with no hydrocephalus. The baby was prepared for elective spinal surgery. Intra operatively , detethering was done, spinal defect repair was checked for any CSF leak. Post operatively the baby was doing well and he was discharged on day 14 of operation.

Fig -15- picture showing sacral cleft with tethering of cords.

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Discussion-

The incidence of NTDs is 0.5–2/1000 live births worldwide. 60%–70% of affected children were females, with craniorachischisis involving cervicothoracic region of the spinal cord.[4] Neural tube development and closure occurs during the 3rd and 4th gestational weeks, and closure of neural tube is completed 28 days postconception.[6] Gene mutation of folic acid metabolism is one of the risk factors for NTDs, although dietary and environmental factors cannot be ruled out Other midline defects involving diaphragm, cleft palate, and cleft lip can also be seen.[7] Many epidemiological studies have suggested that folic acid

supplementation is associated with lesser incidence of NTDs and also associated anomalies such as omphalocele.[6,8] Other anomalies known to be associated with craniorachischisis are cardiovascular defects, caudal regression syndrome or syringomyelia, hypoplastic lung, and gastrointestinal atresia.[9] NTD has occurred despite folic acid supplementation.

Craniorachischisis was seen associated with a mediastinal bronchogenic cyst in a consanguineously married couple, in a case report by Prashant et al.[14] Consanguinity has been implicated in increased cases of NTDs in several countries.[2] Among genetic factors, processes regulating one-carbon folate metabolism and planar cell polarity are strongly considered to cause NTDs.[7,9] MTHFR gene mutations C677T and A1298C are well known to be associated with increased risk for NTDs in humans. A case–control study in the Netherlands by van der Put et al. reported a mutation in C677T in 15% of spina bifida patients and also in the parents of NTD patients. The C677T mutation/polymorphism of the MTHFR gene affects the homocysteine levels in the serum by altering the functional activity of the MTHFR enzyme.[10] The concomitant occurrence of NTDs with omphalocele was also associated with C677T mutation of MTHFR gene.[11] According to a study by Padmanabhan, twinning is a significant factor for the development of NTDs, since more cases of NTDs are seen in families with twins. Monozygotic twins are more prone to congenital anomalies than dizygotic twins.[11] Prenatal counseling, antenatal care and perinatal folic acid

supplementation, and fortification coupled with early screening are some of the effective preventive measures.[12] Over the past three decades, a significant decline in NTD births has been recognized.[7]

Conclusion-

Neural tube defects are owing to persistent non-closure or reopening after the closure of the neural tube. . Failure of primary neurulation leads to ‘Open’ NTDs as comprehended in anencephaly, myelomeningocele (open spina bifida), and craniorachischisis. Failure of secondary neurulation leads to ‘Closed’ NTDs, covered with overlying epithelium, resulting in either asymptomatic spina bifida occulta or severe spinal cord tethering. . The embryonic development of NTDs is multifaceted, with various cellular and molecular mechanisms functioning at different levels of the body axis. Folate supplementation before conception and continuing so throughout pregnancy is beneficial in preventing

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NTDs by 70%. Rest is conferred to as the folate-resistant NTDs. Pre-conception maintenance of adequate BMI should also be suggested. Surgical correction is necessary for spinal NTDs (spina bifida, myelomeningocele) to prevent neurological handicaps. This is a case series ,first of its kind that has been conducted in Tripura Medical College & DR.BRAM teaching hospital, which not only proved the development in the infra-structure of the institute but also the efficacy of multidisciplinary approach in dealing such complicated cases successfully.

Acknowledgment-

The author sincerely thank those who participated in the present study with their own consent. Results from such studies can potentially increase humankind's overall knowledge that can then improve patient care. Therefore, these babies and their families deserve our highest gratitude.[13] Author also thank the Department of Pediatrics, for providing the post-operative supportive care for our study.

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