**TITLE**: **A CASE REPORT OF CONGENITAL HYDROCEPHALUS WITH MYELOSCHYSIS**

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**Introduction**

Neural tube defect (NTD) is a result of dysfunction in neurulation process between third and fourth week of gestation. Myelomeningocele (MMC) is the most common NTD. It is an open spinal cord defect that protrudes dorsally, is not covered by skin, and is usually associated with spinal nerve paralysis. This can lead to a sensory function loss, lower limb paralysis, bladder and bowel dysfunction. Incidence of spina bifida is 1 of 1000 new-born’s1. The aetiology is multifactorial, starting with genetic mutations to an inadequate nutrition2.

Hydrocephalus is a pathologic increase in intracranial cerebrospinal fluid (CSF) volume, whether intraparenchymal or extra parenchymal, independent of hydrostatic or barometric pressure23. Hydrocephalus may result from either fluid production that exceeds absorption or due to mechanical obstruction to the flow of CSF at some level. The site of obstruction may be inside the ventricular system (noncommunicating or internal hydrocephalus) or outside the ventricles (communicating or external hydrocephalus)2. The incidence of a congenital hydrocephalus is 2-3 of 5000 pregnancies.

Folic acid deficiency leads to higher risk of NTD. Folic acid supplementation before getting pregnant reduces incidence of NTD by 71-90% and during the pregnancy period by 70% 3. Folic acid supplementation is highly recommended to be 0.4 mg/day until 12th week of pregnancy for low risk women and for high risk pregnancies (diabetes mellitus, obesity, family history with NTD’s, epilepsy etc.) to be 1-5 mg/day4.

**Case Report**

A 25 year old third gravida with history of previous 2 vaginal deliveries, she was referred to antenatal OPD of AIIMS, Raipur at 34 week of pregnancy for further management.

Her Ultrasonography (USG) examination at 28 weeks revealed gross hydrocephalus (dilated lateral and third ventricle), posterior fossa is obscured by grossly dilated supratentorial ventricular system. Lower lumbar spine and sacral segment show absent posterior element, suggestive of foetal open spina bifida with tethered cord and polyhydramnios. Foetus biometry - BPD: 10.39 cm; HC: 37.93 cm; AC: 26.16 cm; FL: 6.07 cm. Estimated weight: 2092gm; AFI: 21cc; Deepest vertical pocket=9.37cms. Placental attachment–Anterior grade I, upper mid uterine segment. HR 145 b/min. High Impedance flow in both the uterine artery, Mean PI-1.3(90 percentile). Umbilical Artery PI-1.4.MCA show normal flow CPA IS 2.3.



Fig2.Lower lumbar spine and sacral segment show absent posterior element, suggestive of foetal open spina bifida with tethered cord



Fig1.Ultrasonography (USG) examination at 28 weeks revealed gross hydrocephalus

Obstetric history with an intra uterine death at 7month in 2017 and another pregnancy with neonatal death on day 3 due to meconium aspiration syndrome in 2018, both delivered vaginally. Her Past, personal and family history was uneventful.

MRI was planned after 1 week as advised by neonatologist and paediatric surgeon.

She came to labour room with preterm labour on 25/1/20 at 35 weeks . Emergency LSCS was done and a preterm, alive, girl of 2.26kg with gross hydrocephalus and Myeloschysis(ruptured meningomyelocele) was delivered. Head circumference was 53 cm. Apgar scores is 7/10 and 8/10..Baby was shifted to NICU for further management . Myelomeningocele in lumbosacral part was covered with thin layer of sterile dressing. Neurological evaluation revealed decreased tone of the lower-extremities .





USG Cranium- Prominence of the massa intermedia and beaked tectum with dilated lateral and third ventricle suggestive of gross hydrocephalus and Both cerebral hemisphere parenchyma thickness 13 mm.

CECT was done - Tectal beaking, cerebellar tissue wrapping around the brainstem with fenestrations of the falx suggestive of hydrocephalus. Deep scalloping between the bony septations with the lacunar skull (luckenschadel) abnormally large foramen magnum and the flat floor of the posterior fossa. Heart-shaped tentorial incisura that appears to be completely plugged with the upwardly herniating cerebellum. The cerebellar hemispheres extend anteromedially and almost completely engulf the brainstem suggestive of Chiari II malformation

2D ECHO and USG whole abdomen and KUB was normal. After the birth neurosurgeon’s consultation was provided

**Discussion**

Patient had emergency caesarean as she came in preterm labour and did not consent for. Prognosis of the baby was explained to the couple.

Hydrocephalus without an obvious extrinsic cause in infants is usually referred to as congenital hydrocephalus, since it is often present at birth.

Most common causes of congenital hydrocephalus include aqueduct stenosis, meningomyelocele (chiari malformation), intrauterine infection (CMV, toxoplasmosis, syphilis), agenesis of corpus callosum, X-linked hydrocephalus syndromes, intracranial haemorrhage, Dandy–Walker malformation, and intracranial tumours6.

In this case of Hydrocephalus with myelomeningocele with most probable aetiology is Arnold Chiari type II malformation as seen in post natal CECT.

The majority of patients with myelomeningocele has hydrocephalus. In this setting, hydrocephalus is caused by the Chiari II malformation which obstructs outflow of CSF from the fourth ventricle and/or flow through the posterior fossa. In addition, there is often associated aqueductal stenosis. Hydrocephalus associated with myelomeningocele tends to have both an obstructive component and a communicating component 7.

The Chiari II malformation seen in spina bifida is acquired and is accompanied by other features on MRI, such as agenesis of corpus callosum, low lying torcular herophili, tectal breaking, medullary kinking, and heterotopias 8.

Isolated hydrocephalus is frequently caused by aqueductal stenosis8.

The hereditary nature of hydrocephalus was first appreciated by Bickers and Adams in 1949. It is now known that mutations in L1CAM, which result in a collection of X-linked conditions known as the L1 spectrum disorders, account for the majority of inherited cases of hydrocephalus10. The L1CAM gene product is a neural recognition molecule that plays key roles in neuronal migration and axon guidance. When mutated, it gives rises to aquaductal stenosis, isolated agenesis of the corpus callosum and X-linked spastic paraplegia6.

Fried syndrome, is caused by mutations in the AP1S2 gene6.Trisomy 13, 18, 9 and 9p can also present with hydrocephalus11.

Intrauterine infections such as rubella, cytomegalovirus, toxoplasmosis, lymphocytic choriomeningitis, syphilis, and Zika virus can result in congenital hydrocephalus. 11.

For families with no other family history and negative L1CAM mutation testing, the recurrence rate of hydrocephalus is 4% for couple with previous child with hydrocephalus12.

In the middle of the 4th week, neural folds on each side of the neural groove begin to fuse, thus forming the neural tube20. At the level of the fifth somite, where the brain and spinal cord meet, the normal folds join in a zipperlike fashion that proceeds cranially and candally21. A second closure site appears in the forebrain; fusion also occurs at that site in two directions, and meets the zipper process proceeding from the hindbrain. In parallel, the zipper process moves to close the most rostral part of the forebrain failing which lead to NTD. 22

Diagnosis of hydrocephalus, can be determined in utero by USG as early as 12 weeks of gestation or later with MRI. Postnatally USG is helpful, brain MRI remains the best diagnostic test 17. One of the laboratory methods to suspect NTD is maternal serum alpha-fetoprotein (AFP) testing in a triple screen or quadruple screen test 18.In this case it was diagnosed late since, Antenatal follow up was not proper .she had not done quadraple marker test and 1st her usg was done at 28 weeks of gestation.

Following are the procedures available for CSF reduction ventriculoperitoneal shunt operation ,choroid plexus coagulation , cephalocentesis13 for hydrocephalus. Repair surgeries available for MMC are In-utero fetal surgery11, mini-hysterotomy24,skin closureand amniotic membranes (AM) repair16. The Management of Myelomeningocele Study (MOMS) trial showed a significant reduction of ventriculoperitoneal shunt placement at one year of age following foetal surgery (prenatal group: 40%; postnatal group: 82%) 11

Nevertheless, foetal surgery was associated with significant risks related to premature birth, thinning of the uterine wound, tissue edge separation at the hysterectomy site, oligohydramnios, chorioamniotic separation, placental abruption, and spontaneous membrane rupture 11.

Recent advance of Fetal MMC repair is feasible through a mini-hysterotomy . This approach appears to be associated with reduced risks of very preterm delivery and maternal, fetal and neonatal complications as compare to open repair24.

The external ventricular drain was inserted on the second day of life in this case . After two weeks the external ventricular drain was replaced with ventriculoperitoneal shunt. The baby was discharged one month after replacement of ventriculoperitoneal shunt. Advised neurosurgeons and neonatologist follow up once in a month. VP shunt is required in children with increased HC (≥ 2 standard deviation regarding age group), bulging fontanels, or LVA width of ≥ 15 mm after the closure of MMC 14. It has Abdominal complications include intestinal obstruction, volvulus, peritonitis, peritoneal cyst, CSF ascites15.

**Conclusion**

We have reported a case of a new born with severe hydrocephalus and myeloschiasis. Multidisciplinary medical team (obstetricians, neonatologists, paediatric surgeon and neurosurgeon) were working in close relations.  Early antenatal diagnosis of neural tube defects can provide the best child delivery strategy. Multidisciplinary post-delivery care of new born including follow-up of vital functions and adequate neurosurgical care should be provided.

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