A case report of Arnold Chiari Malformation Type II with Cervical Myelomeningocele and multiple congenital malformations

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Introduction

Definition and classification

- Arnold-Chiari Malformation is defined as a group of deformities of the hindbrain (cerebellum, Pons, and Medulla oblongata), it could be anything from cerebellar herniation to the absence of the cerebellum itself, sometimes associated with other neurological malformation such as hydrocephalus, encephalocele and myelomeningocele.
- Chiari malformations are classified into four types based on their morphology and the severity of anatomical defects. [1]

The Chiari II Malformation

- The Chiari II Malformation is relatively common congenital malformation with an incidence of about 1 in 1000 live births, Characterized by herniation of the cerebellum in addition to the herniated cerebellar tonsils and vermis through the foramen magnum into the spinal canal due to an open distal myelomeningocele.

- Nearly all the patients who had myelomeningocele have Chiari II Malformation.

Etiology

- The etiology in most cases of myelomeningocele is multifactorial, involving genetic, racial, and environmental factors, in which nutrition, particularly folic acid intake, is key.
- Most infants born with myelomeningocele are born to mothers with no previously affected children.
- Up to 10% of fetuses with a neural tube defect detected in early gestation have an associated chromosome abnormality.
- In women with pregestational diabetes, the risk of having a child with a CNS malformation, including myelomeningocele, is 2-10 fold higher than the risk in the general population.

Myelomeningocele

- Myelomeningocele (MMC), is a spina bifida in which the spinal cord and its contents herniate through a congenital bony deformity of the spine.[4]
- **Cervical myelomeningocele** (CMMC) is an uncommon congenital anomaly of the spinal cord and represents only 1 – 5 % of all neural tube defects. CMMC presents with different clinical characteristics compared with the other more common lumbosacral type.[5]
- **ORPHA:93969**

Case Presentation

- A male neonate was admitted to pediatric department on day one post-delivery because of myelomeningocele in his cervical region, hydrocephalus and multiple congenital anomalies.
- He was born at 39 weeks of gestation by emergency transverse cesarean section.
- His birth weight was 4700 g.
- He was admitted to neonatal ICU with respiratory distress which resolved in the first week.

Case Presentation-Examination

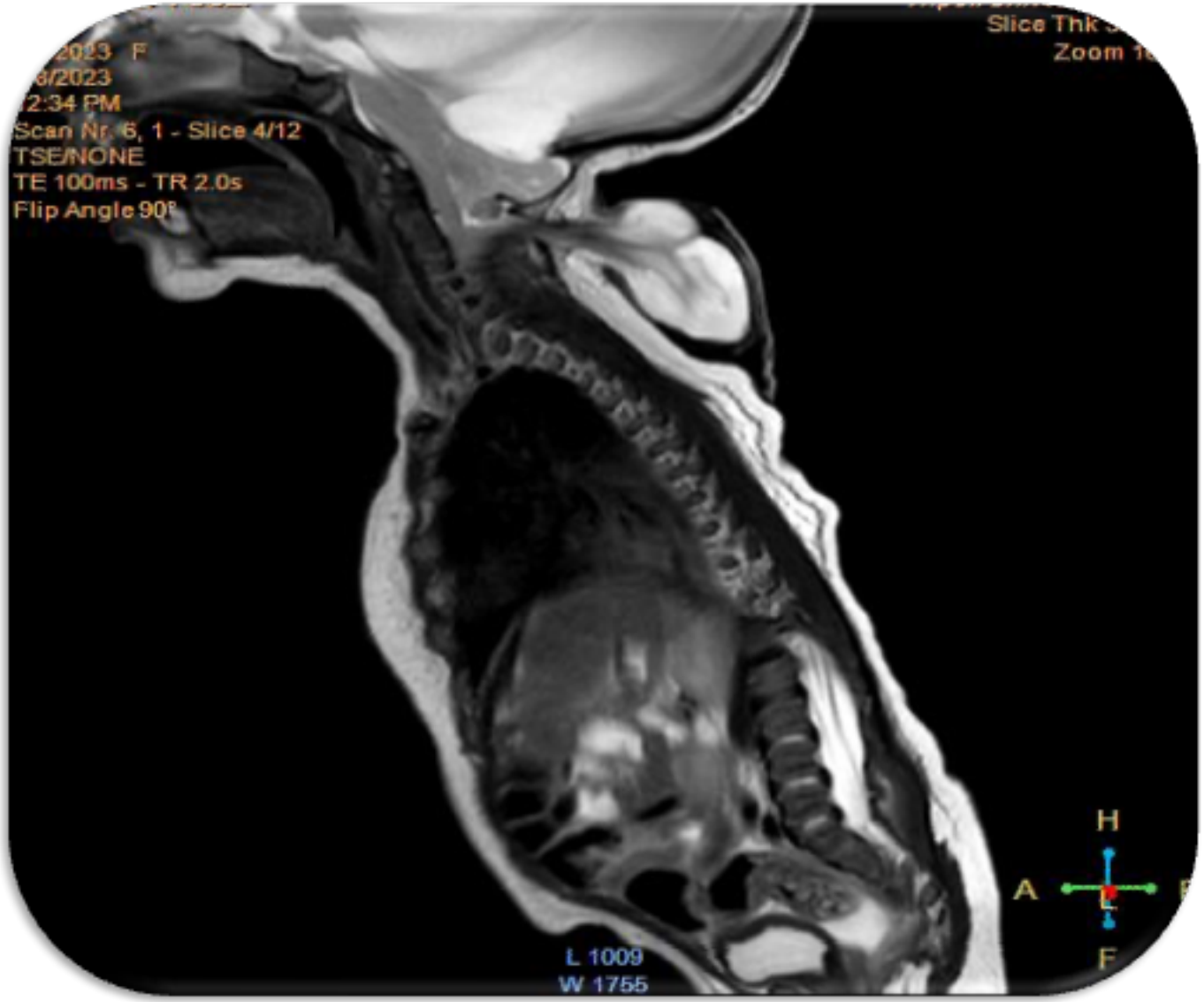
- On examination the baby looks slightly distressed, anterior Fontanelle is elevated, the occipeto-frontal circumference was 49 cm.
- Moro's reflex, suckling reflex, and papillary reflex were positive. On the other hand the gag reflex was significantly decreased. On examining the upper limp both limbs revealed hyporeflexia, much more greater on the left side. Tone and sensation were negative on both limbs, the left shoulder showed an abduction of 130^{o}, while the right shoulder abduction degree was 150^{o}.
- The lower limb examination shows a full abduction of the hip joint, 120^{o} flexion of both knees. The planter reflex was positive and sensation was significantly present.
- In brief weakness appeared more on the upper limb than the lower limb, and the left side more than the right side.

Case Presentation- Imaging

- Magnetic resonance imaging (MRI) revealed dilatation of 3rd ventricles, compression of the 4th ventricle, downward displacement of the midbrain into the spinal canal, and myelomeningocele at C2. The case was diagnosed as Arnold-Chiari malformation type II with cervical myelomeningocele and obstructive Hydrocephalus.

- Echocardiography showed a small patent foramen oval.

Advanced Imaging techniques



MRI here shows cervical myelomeningocele at second cervical vertebra, with syringomyelia

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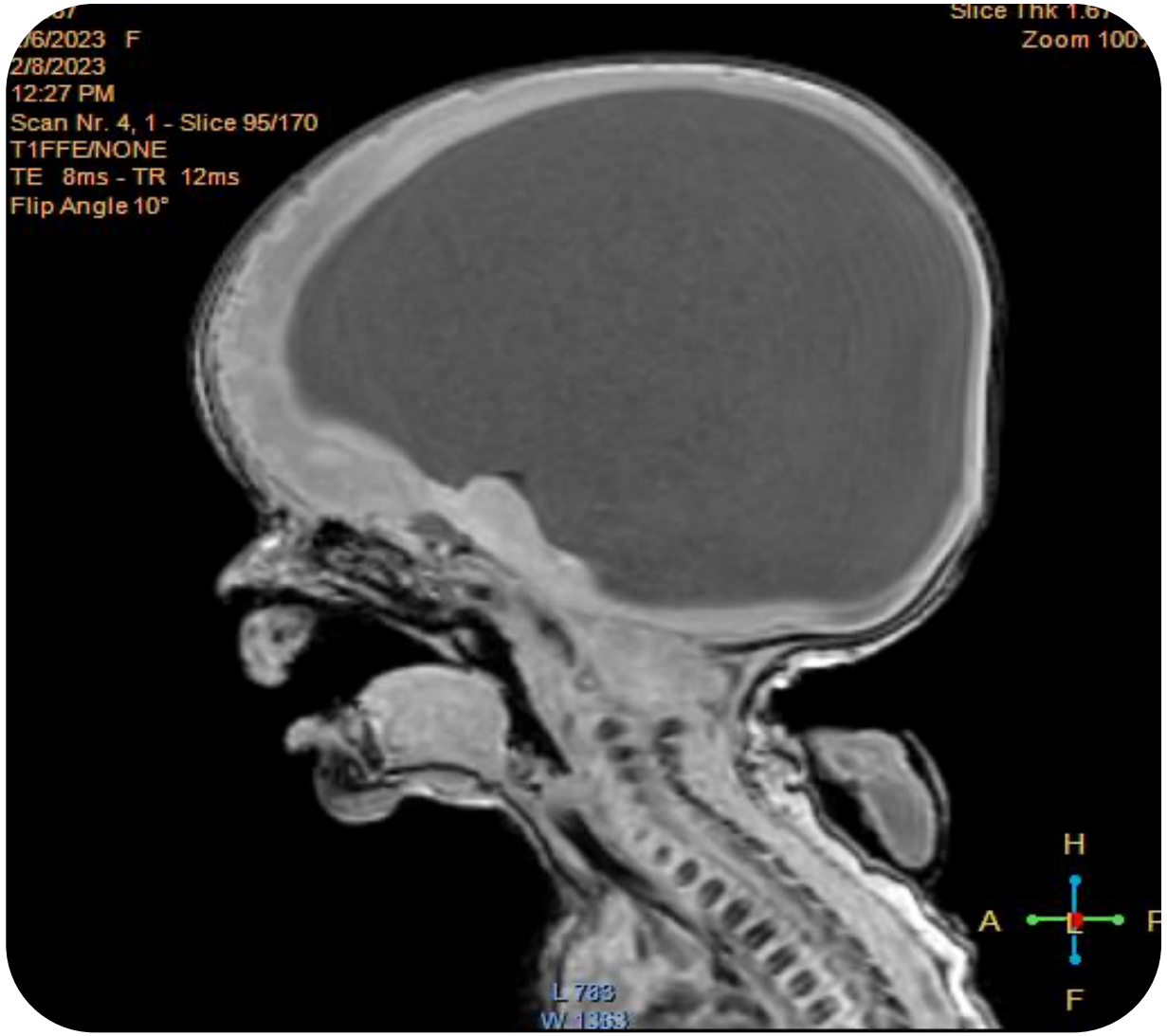
Advanced Imaging techniques



This MRI image shows downward displacement of the midbrain and blockage of the spinal canal.

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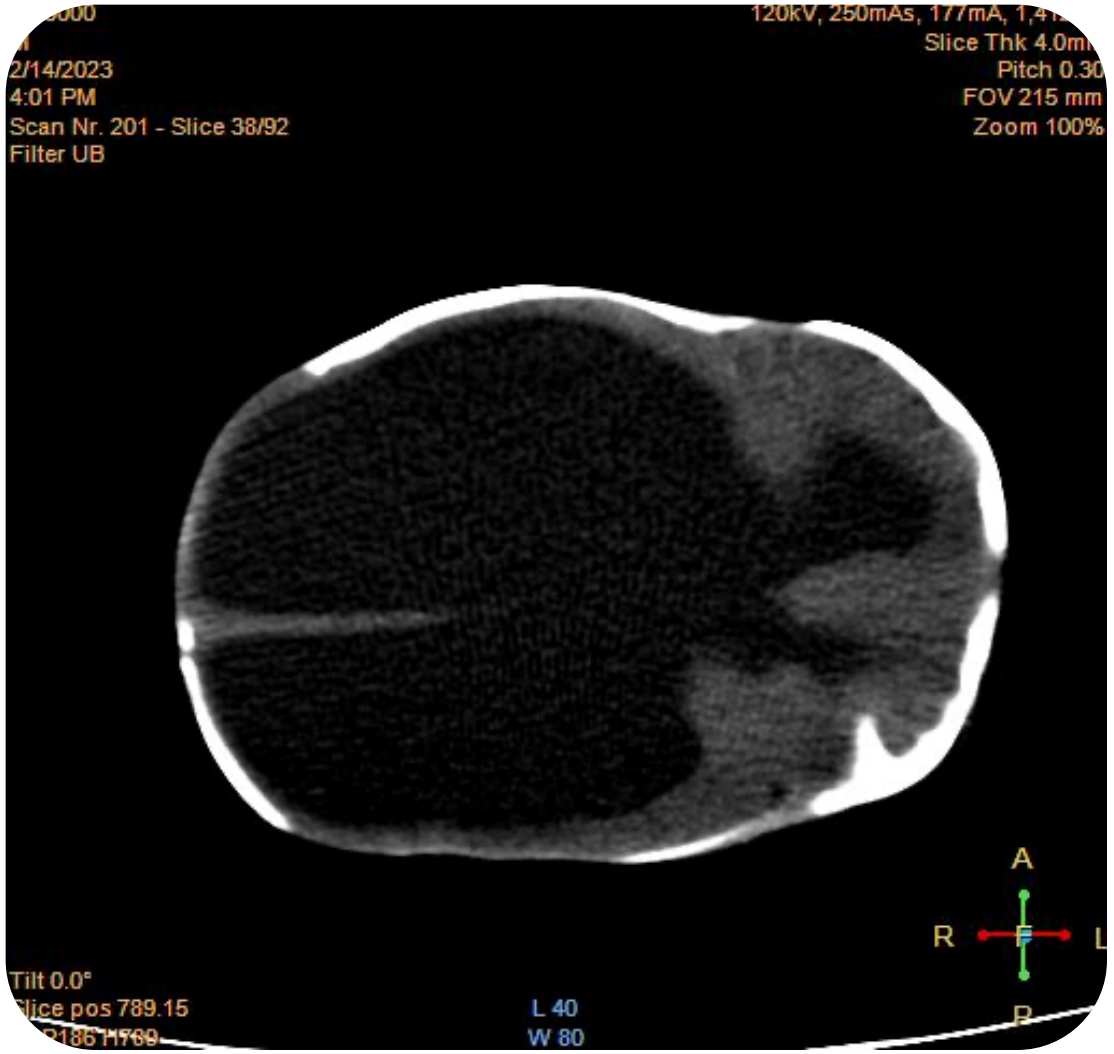
Advanced Imaging techniques:



CT-Scan shows hydrocephalus compressing the brain tissue.

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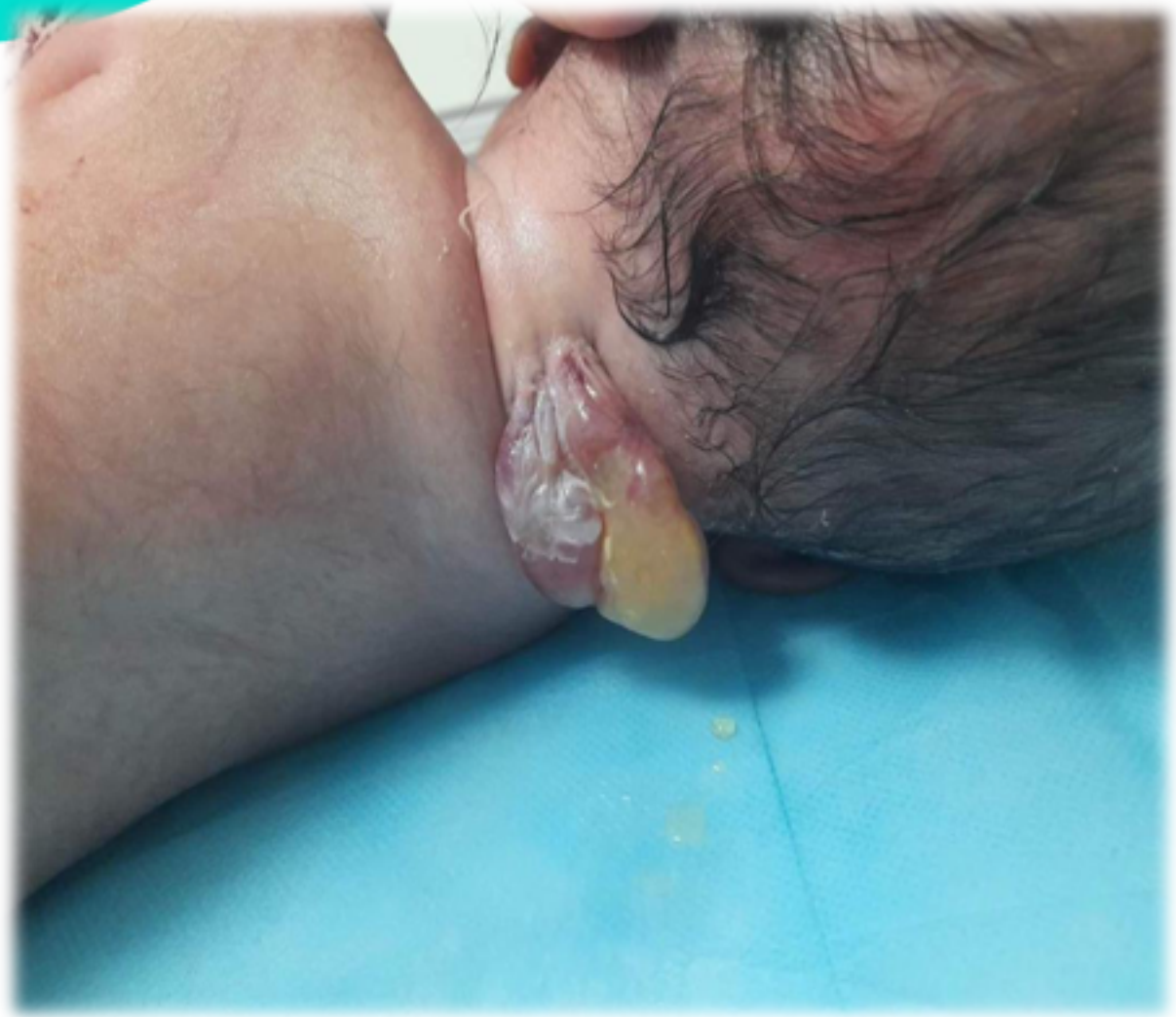
Cervical Myelomeningocele with hydrocephalus



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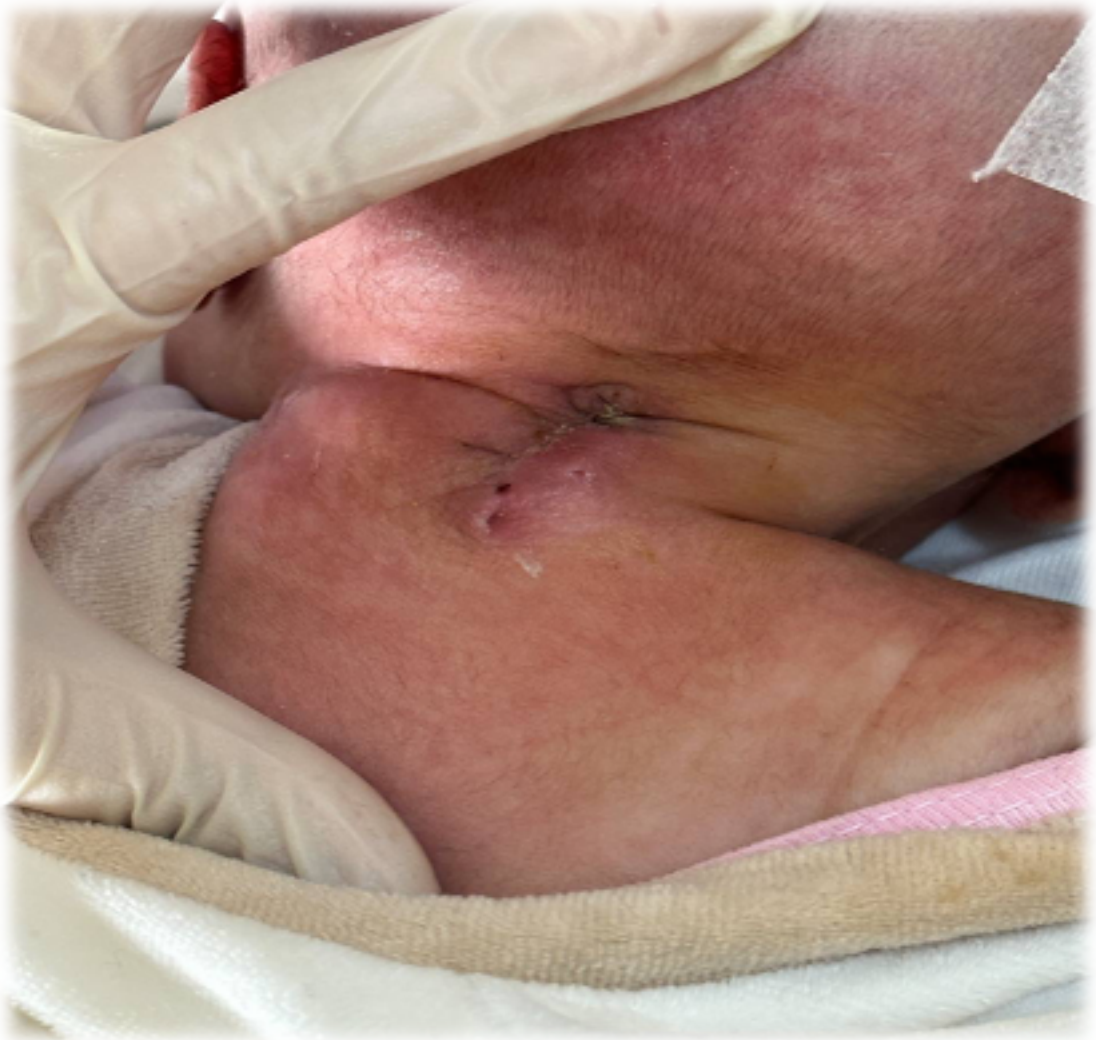

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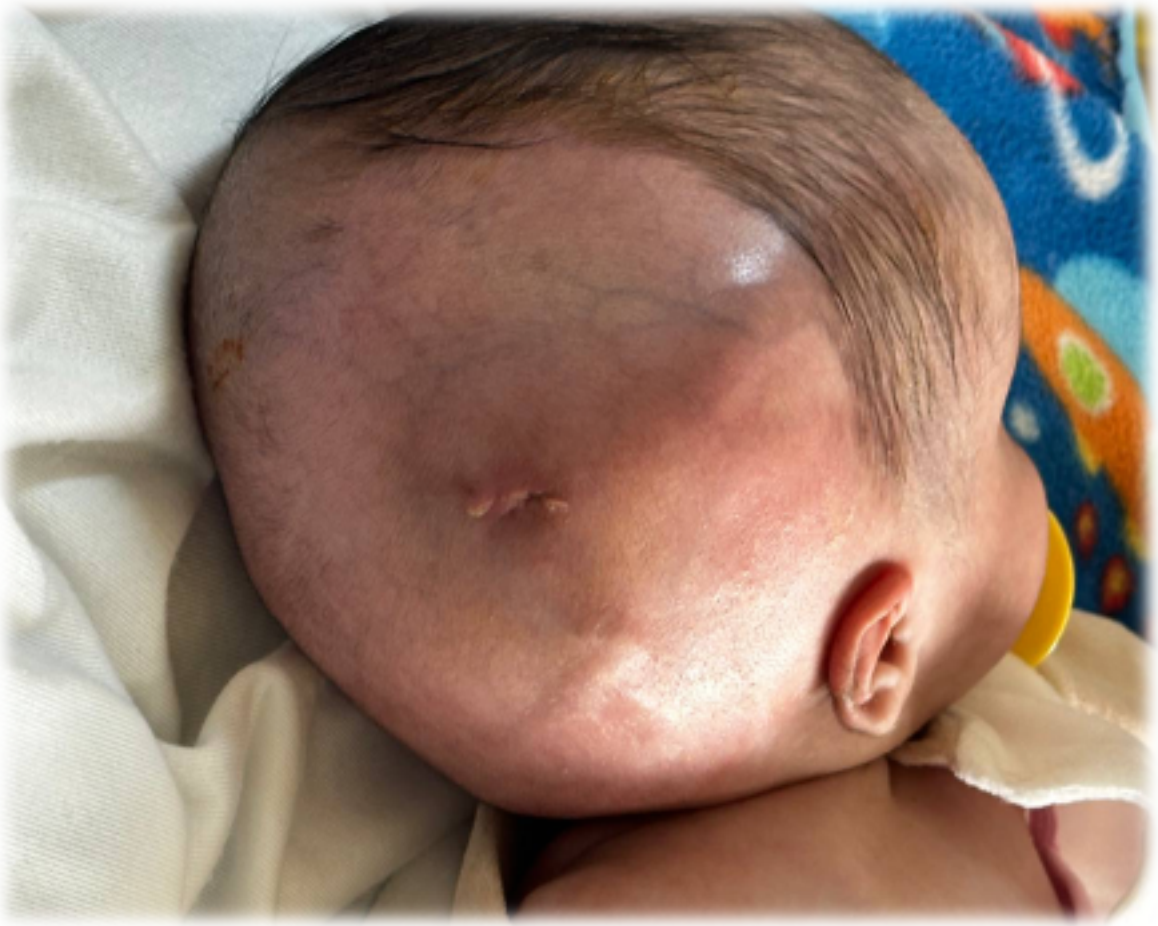

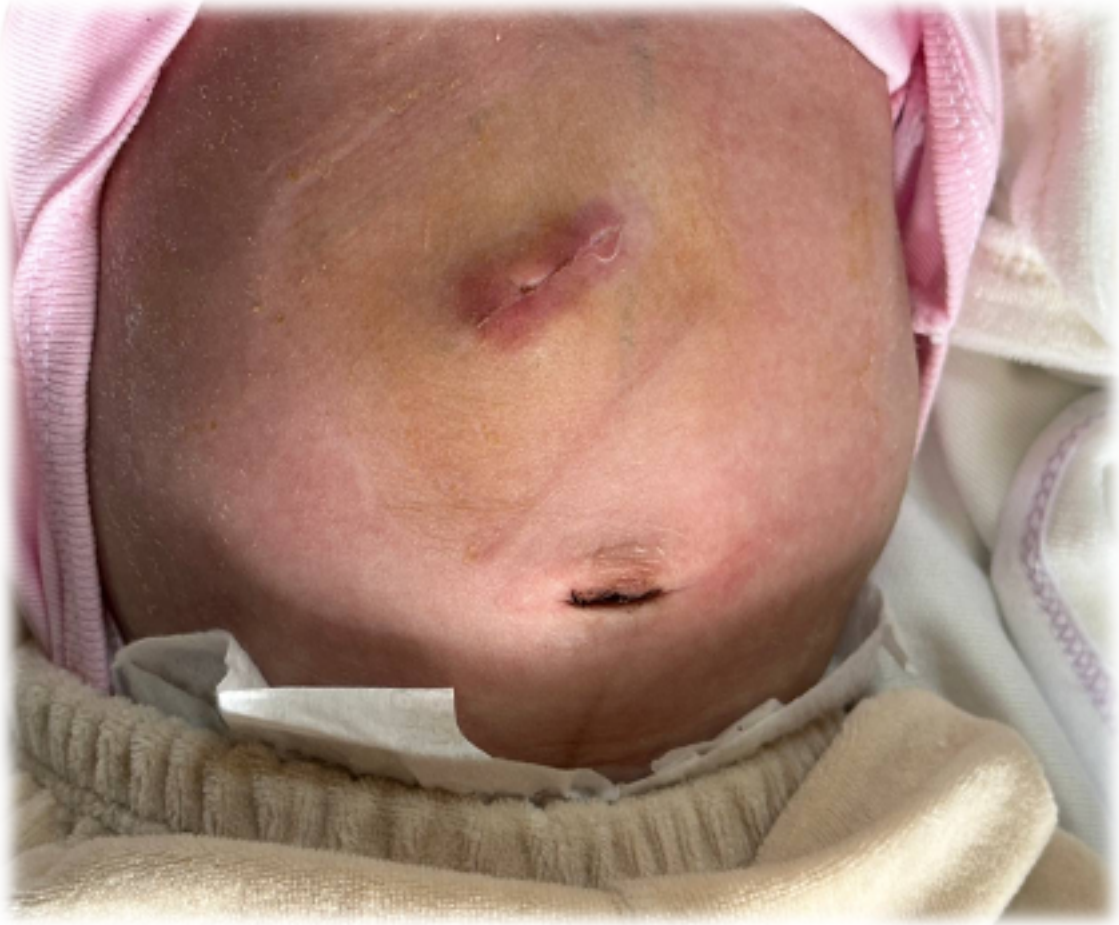

Ventriculo-peritoneal (VP) shunt

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Case Presentation- Management

- On the 11th day post delivery a surgical intervention was performed by prof. Dr. Faisal Ben Taleb in Neurosurgery department at Tripoli University Hospital.
- The procedure involved repairing the cervical myelomeningocele and inserting a Ventriculo-peritoneal shunt. The myelomeningocele was repositioned inside the spinal cord, and a dura was created and closed with no cerebrospinal fluid leakage.
- The wounds were dressed, and the baby was kept under observation for four days. Afterward, significant improvement was noticed in the baby's respiratory distress, and the anterior fontanelle was at level. On the 5th day post-operation, the baby was discharged home and advised to follow up with the neurosurgical department after one week.

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Case Discussion

- The report highlights the importance of timely intervention in rare and complex conditions
- In this case, the baby presented with respiratory distress, which is a common complication of cervical myelomeningocele due to the involvement of the upper spinal cord segments that control respiration.
- The distress improves after surgical intervention.
- Cervical myelomeningocele can be associated with other conditions, such as Chiari malformation, this can lead to hindbrain compression and a range of neurological symptoms, including headaches, dizziness, and motor and sensory deficits. Chiari malformation can also cause respiratory distress.

Case Discussion- complications

- Manifestations of brainstem dysfunction caused by hindbrain hernia and aggravated by ventricular dilatation include the following:

1. Poor feeding, Poor sucking and Recurrent vomiting
2. Generally quiet behavior with poor crying
3. High-pitched cry or stridor caused by vocal cord paralysis - A predictor of poor outcome
4. Episodes of apnea
5. Extremity weakness in older children
6. Recurrent aspiration - Often manifesting as recurrent pneumonia

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Prognosis

- In a 20-year follow-up survey of children who received shunting in the 1970s, more than half of them graduated from mainstream education.
- In a review of a cohort of patients treated in the 1970s for spina bifida aperta, 52% of the patients were alive 20 years after treatment. Most of the deaths occurred in the first year of life, mostly due to renal and respiratory problems associated with spina bifida.
- In a review of children treated in the 1980s, only 27% died; most of them died in the first year of life from causes related to spina bifida rather than hydrocephalus.
- Approximately 75% of children with myelomeningocele have an IQ higher than 80. Among those whose intelligence is normal, 60% are learning disabled.

Case Discussion

- The The Management of Myelomeningocele (MOMS) trial completed in 2011 found that repairing myelomeningocele (MMC) before birth, in the uterus, can improve outcomes for babies with MMC. Prenatal treatment can help reduce or eliminate complications such as hydrocephalus, Chiari malformation, and lack of movement in the lower extremities. Early diagnosis and repair of MMC can have a significant impact on the baby's development.
- The overall incidence of myelomeningocele has significantly declined in the past two decades because of improved maternal nutrition during pregnancy, including the addition of folic acid, a wider availability of antenatal diagnosis, and therapeutic termination of pregnancy.

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Conclusion and recommendations

- The case highlights the importance of multidisciplinary management and timely intervention in rare and complex conditions, such as cervical myelomeningocele and Chiari malformation.
- To further reduce the risk of neural tube defects, it is important to raise awareness among women of childbearing age about the importance of taking folic acid supplements before becoming pregnant. Public health campaigns and education programs can play a critical role in promoting this message and helping to ensure that women have access to the information and resources they need to make informed decisions about their health.
- Early detection and diagnosis of myelomeningocele is crucial for optimal outcomes. Ultrasound screening in early pregnancy can help identify this and other congenital anomalies, allowing for timely intervention and management. Healthcare providers should encourage all pregnant women to undergo routine prenatal screening, including ultrasound, to identify any potential issues early in the pregnancy.

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Thank You


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