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Chiari Malformation: A Case Study

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Abstract: Chiari malformation comprises a heterogeneous group of congenital anomalies involving the cerebellum, brainstem, and craniocervical junction, characterized by caudal herniation of cerebellar structures, with or without involvement of the medulla, through the foramen magnum into the spinal canal. These defects are associated with significant perinatal morbidity and mortality, contributing to the burden of congenital anomalies in the community. Early identification through systematic antenatal screening is essential to ensure timely referral, appropriate counselling and continuity of care. Routine prenatal ultrasonography remains a primary, non-invasive modality for early detection, particularly in resource-limited settings where community health nurses play a pivotal role in antenatal surveillance, risk identification and health education. This case report describes a primigravida at 22 weeks of gestation who presented to an antenatal clinic, where ultrasonography revealed findings suggestive of fetal Chiari malformation. The case underscores the importance of strengthening community-based antenatal services, improving awareness among expectant mothers, and enhancing the role of nursing professionals in early diagnosis and referral systems to reduce adverse fetal outcomes.

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I. INTRODUCTION

Arnold–Chiari malformation was initially described in 1891 by the Austrian pathologist Hans Chiari during the examination of pediatric autopsy specimens. The disorder was later named in recognition of both Chiari and his mentor Julius Arnold.

Chiari malformation is a congenital hindbrain anomaly. It is commonly classified into types I and II, with Type II frequently associated with neural tube defects such as myelomeningocele. These abnormalities may result in significant neurological complications and perinatal morbidity. Prenatal diagnosis through ultrasonography, using markers like the lemon and banana signs, facilitates early detection. Emphasis on antenatal screening, early referral, maternal education, and folic acid supplementation is essential for prevention and improved pregnancy outcomes.

II. CASE HISTORY

A 19-year-old pregnant woman attended a routine antenatal clinic at S.S. Kulam Community Health Center, Coimbatore, Tamil Nadu. She was a primigravida and had been married for one year in a non-consanguineous marriage. Her menstrual cycles were regular. The last menstrual period was recorded as 02.03.2025, and the expected date of delivery was estimated as 09.12.2025.

The patient had no history of contraceptive use or intake of ovulation-inducing medications. Clinical examination revealed moderate pallor. She was receiving iron and folic acid supplementation, and her haemoglobin level was 9 g/dL. Fetal movements were present and the fetal heart sounds were clearly audible. During the course of pregnancy, she was diagnosed with hypothyroidism and was prescribed Tab. Thyroxin 50 µg once daily. A routine ultrasound scan performed at 22 weeks of gestation identified multiple fetal anomalies. These included mild bilateral ventriculomegaly with a dangling choroid plexus. The ultrasound findings also showed an abnormal skull configuration known as the lemon sign, an altered cerebellar shape referred to as the banana sign, and spina bifida with myelomeningocele in the thoracolumbar region.

Based on these sonographic findings, a diagnosis of Arnold–Chiari Malformation Type II was made. After counselling the couple regarding the condition and prognosis, termination of pregnancy was advised. Following informed consent, the mother was referred to Coimbatore Medical College Hospital for further management.

➢ *Prevalence*

The estimated prevalence of Chiari malformation in the general population is slightly less than 1 in 1000 individuals.

- Type I is the most frequently reported form, occurring in approximately 1 out of every 1000 births. Studies also

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indicate a slightly higher occurrence among females, with a ratio of approximately 1.3:1.
- Type II Chiari malformation is strongly associated with neural tube defects, particularly myelomeningocele.

➢ *Causes*

When Chiari malformation develops during fetal life, it is referred to as primary or congenital Chiari malformation. Possible contributing factors include:

- Genetic mutations affecting normal brain development
- Inadequate maternal intake of essential nutrients, especially folic acid
- Maternal infections or high fever during pregnancy
- Exposure to harmful chemicals, alcohol, or illicit drugs during gestation

Certain medications, such as selective serotonin reuptake inhibitors (SSRIs) used for the treatment of depression during pregnancy, have also been reported to increase the risk of Chiari malformation in the fetus.

Secondary or acquired Chiari malformation may occur later in life when cerebrospinal fluid is drained excessively from the lumbar or thoracic region of the spine. This may result from traumatic injury, disease, or infection.

➢ *Types of Chiari Malformation*

Chiari malformation is classified into four main types based on severity and anatomical features.

- Type I: Characterized by herniation of the cerebellar tonsils without association with myelomeningocele.
- Type II: Involves herniation of the cerebellar vermis and brainstem into the spinal canal. It is commonly associated with spina bifida and features such as hydrocephalus, medullary kink, and tentorial dysplasia.
- Type III: A rare and severe form involving herniation of brain tissue associated with cephalocele or cranio-cervical meningocele.
- Type IV: Marked by severe cerebellar hypoplasia and downward displacement of posterior cranial fossa contents.

Additional variants have also been described, including Chiari Type 0 and Chiari Type 1.5.

➢ *Diagnosis*

Prenatal diagnosis of fetal anomalies is primarily performed through routine ultrasonographic examination during the second trimester of pregnancy. This screening method can identify approximately 70–90% of major congenital abnormalities.

In cases of Chiari malformation, characteristic sonographic markers are often observed. Two well-recognized findings include frontal bone scalloping, commonly referred to as the lemon sign, and an abnormal curvature of the cerebellum known as the banana sign. These findings suggest the presence of neural tube defects and associated hindbrain abnormalities. (Ref.Fig.1)

With advancements in prenatal imaging, these structural changes may be detected at an early stage of pregnancy. The lemon sign and banana sign can sometimes be identified as early as 12 weeks of gestation. Additionally, myelomeningocele may be detected around the 10th week through prenatal imaging techniques.

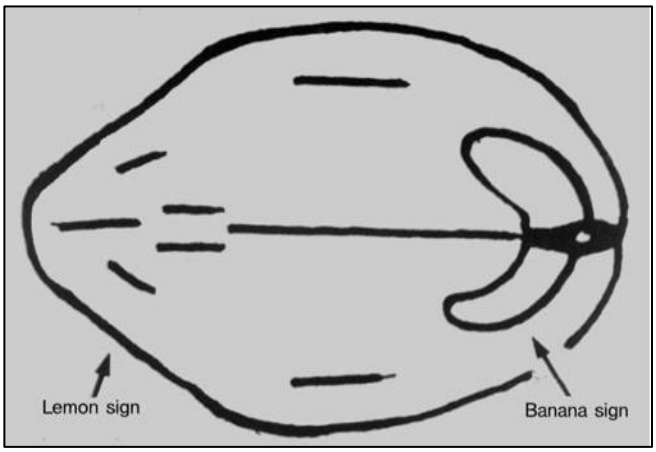

Fig 1 Banana Sign and Lemon Sign

Spina bifida is typically diagnosed by identifying defects in the posterior vertebral elements, such as an open neural arch or widened laminae. In many cases, a fluid-filled sac containing cerebrospinal fluid and neural tissue can be visualized at the site of the defect, which corresponds to myelomeningocele.

➢ *Treatment*

Management options for Chiari malformation diagnosed during pregnancy depend largely on the severity of the condition and the associated anomalies. When severe congenital abnormalities are identified, parents may be offered the option of termination of pregnancy after appropriate counselling and medical evaluation.

If the couple choose to continue the pregnancy, specialized intrauterine interventions or advanced prenatal monitoring may be considered in selected cases. Comprehensive counselling is essential to help couple understand the possible complications, long-term prognosis, and available treatment options.

In addition to medical management, psychological support and guidance should be provided to couple to help them cope with emotional stress and prepare for potential outcomes.

➢ *Prevention*

In many developing countries, including India, a significant proportion of pregnancies are unplanned. Neural tube formation occurs very early during embryonic development, and the rostral and caudal neural pores usually close by the sixth week of gestation. If folic acid

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supplementation is started after this critical period, it may not effectively prevent neural tube defects.

To reduce the risk of congenital anomalies, it is recommended that women of reproductive age consume a daily multivitamin supplement containing 0.4 mg of folic acid and vitamin B12 for at least two to three months prior to conception. Supplementation such as iron and folic acid tablets, calcium tablets should continue throughout pregnancy and for several weeks during the postpartum period which is provided by government sectors under free of cost.

Pregnant women should also avoid exposure to harmful chemicals, alcohol, and illicit drugs, as these substances may interfere with normal fetal development.

Chiari malformation represents an important clinical condition with characteristic imaging findings. Prenatal ultrasonographic screening remains the most effective primary method for detecting early fetal abnormalities. Early detection enables healthcare professionals to guide parents in decision-making regarding further investigations, genetic evaluation, or pregnancy management.

Therefore, strengthening prenatal screening programs and ensuring the availability of diagnostic services at primary healthcare levels are essential for improving maternal and fetal health outcomes.

III. CONCLUSION

Chiari malformation remains a significant congenital condition contributing to adverse perinatal outcomes, emphasizing the need for early detection and coordinated care. Prenatal identification through routine ultrasonography enables timely intervention, appropriate referral, and informed decision-making. From a community health nursing perspective, strengthening antenatal services, promoting early registration of pregnancy, and ensuring regular screening are essential strategies. Community health nurses play a key role in risk assessment, health education, and counseling, particularly regarding the importance of folic acid supplementation and adherence to antenatal visits. Enhancing community awareness and integrating multidisciplinary care can help reduce complications and improve maternal and fetal health outcomes.

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