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Case Report

Antenatally diagnosed myelomeningocele with associated chiari ii malformation in the third trimester. A case report ☆

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ABSTRACT

Myelomeningocele, a severe form of open neural tube defect which is mostly associated with Chiari II malformation remains a cause of adverse neonatal outcomes. Myelomeningocele is mostly detected in the second trimester and subsequently followed up in the third trimester. We present a case of myelomeningocele with associated Chiari II malformation that was diagnosed for the first time in the third trimester in a 32-year-old pregnant woman. Previously performed ultrasound at 12 weeks gestation was unremarkable. Considering the prognosis of the detected anomalies, the patient opted for medical termination of the pregnancy after receiving thorough counselling.

Third trimester anomaly screening should be encouraged as part of routine third trimester scans; particularly in women who report late for antenatal care and those who lack periconceptional folate supplementation.

Introduction

Spina bifida, a form of congenital neural tube defect occurs in approximately 1 per 1000 births; with myelomeningocele being the most severe and clinically significant subtype [1]. Myelomeningocele is characterized by the incomplete closure of the embryonic neural tube during the first few weeks of gestation. This defect eventually results in a sac-like protrusion through the spinal column, which contains meninges, spinal cord, and cerebrospinal fluid (CSF) [2]. Chiari II malformation, almost invariably associated with myelomeningocele, is a congenital anomaly where the cerebellar tonsils and brainstem herniate downward into the cervical spinal canal through the foramen magnum to compensate for the drop in pressure gradient across the foramen magnum due to CSF leakage and drop in intracranial pressure. This displacement occasionally leads to obstructive hydrocephalus due to the blockage of CSF flow at the craniovertebral junction [3,4].

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Although the exact cause of myelomeningocele is not known, several risk factors have been associated with the development of this anomaly. Maternal factors such as lack of periconceptional folate supplementation, smoking, obesity, diabetes; as well as environmental exposure to radiation, teratogenic agents and pollutants such as pesticides and organic solvents have all been strongly linked to the occurrence of myelomeningocele [1,5].

Myelomeningocele with associated Chiari II malformation remains a major cause of severe neurological impairments such as lower limb paralysis, neurogenic bladder, abnormal bowel function and respiratory problems in severe cases; hence early detection with ultrasonography or magnetic resonance imaging (MRI) during the antenatal period is necessary for adequate medical and surgical interventions [6–8].

We therefore present a case of an antenatally diagnosed meningomyelocele with associated Chiari II malformation in a 32-year-old pregnant woman in the third trimester with ultrasound. We further describe the usefulness of third trimester anomaly scan in detecting neural tube defects, particularly in women who lack folate supplementation and in women who present late for antenatal care.

Case report

A 32-year-old woman; gravida 2, para 0 with a previous spontaneous abortion presented at our health facility for a routine obstetric scan at 32 weeks of gestation. This was her first visit to the facility, and she was referred by her attending physician for a detailed scan. The patient revealed that she had not attended regular antenatal care (ANC) after her first trimester scan at a private scan center. She also indicated she had not taken any periconceptional folate supplementation or pregnancy support medications. She had no known prior or current history of hypertension, diabetes or alcoholism.

Investigations and imaging findings

Prior to the current ultrasound, a previous ultrasound scan conducted in a different health facility during the first trimester (12weeks gestation) was unremarkable. The third trimester ultrasound performed at our facility revealed a 3.2 cm defect at the lumbosacral region with herniation of meninges, neural tissue and cerebrospinal fluid (CSF), consistent with myelomeningocele (Fig. 1). Also noted was complete effacement of the cisterna magna with herniation of the cerebellum through the foramen magnum (Fig. 2). Dilated lateral ventricles measuring 12.7mm in diameter and a dilated third ventricle, indicating ventriculomegaly were also detected (Fig. 3). Additionally, polyhydramnios was observed, with an amniotic fluid index (AFI) of 26.6 cm.

These ultrasound findings prompted a diagnosis of myelomeningocele with associated Chiari II malformation.

An MRI and maternal serum alpha-fetoprotein (MSAFP) test were not done because the patient could not afford the costs involved.

Follow-up and treatment outcomes

Even though MRI was not performed, ultrasound performed by experienced sonographers has been seen to be very sensitive in detecting neural tube defects and brain anomalies particularly in the second and third trimesters; hence the diagnosis with ultrasound was enough to allow for clinical decision making. Following the diagnosis, the patient received counseling from her obstetrician. The nature of the diagnosed condition, potential complications, and the likely prognosis were explained to the patient. Furthermore, both medical and surgical interventions such as in-utero surgical repair were discussed with the patient. The potential benefits and risks of each option were discussed in detail, enabling the patient to make an informed decision regarding the management of her pregnancy. The patient opted for medical termination of the pregnancy, considering the disease prognosis and potential impact on the child's quality of life. The chosen management plan involved labor induction.

The patient was administered Mifepristone to block progesterone, necessary for maintaining the pregnancy. After 48 hours, Misoprostol was given intravaginally in 4 doses every 3 hours to soften the cervix and induce uterine contractions.

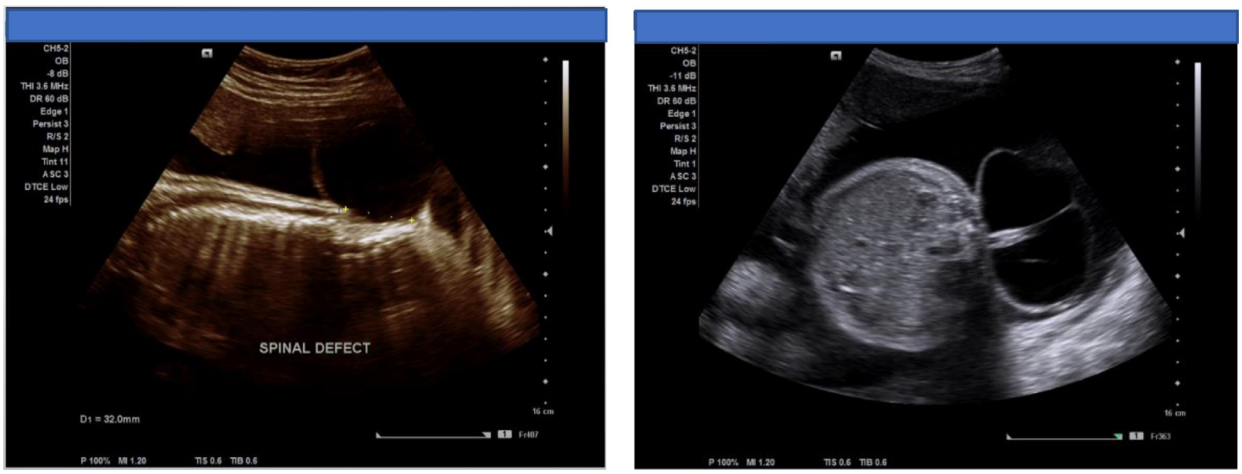

Fig. 1 – Sonograms of the fetal spine (sagittal and transverse view) depicting a 3.2 cm defect at the lumbosacral region with herniation of neural tissues, meninges and accumulation of cerebrospinal fluid in a cystic form.

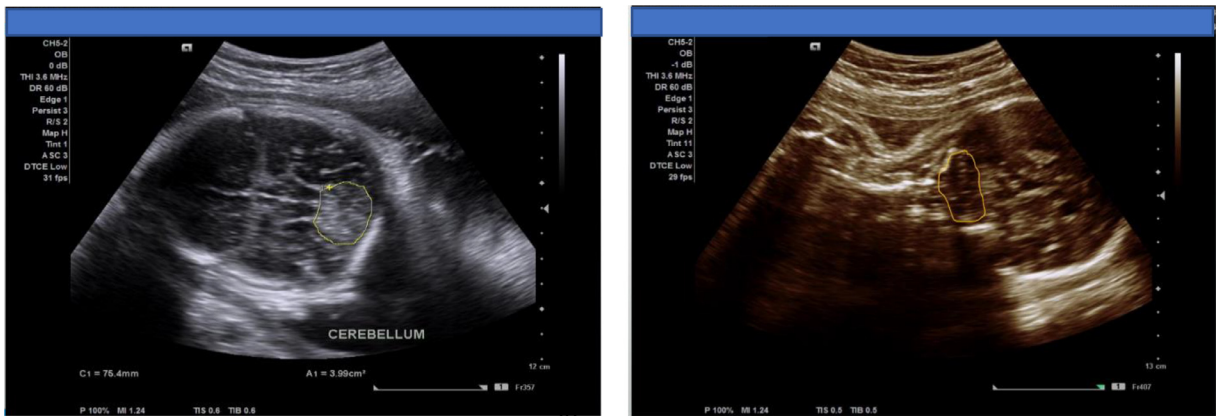

Fig. 2 – Sonogram of the fetal head (axial view) shows complete effacement of the cisterna magna; and downward displacement of the cerebellum through the foramen magnum (coronal view).

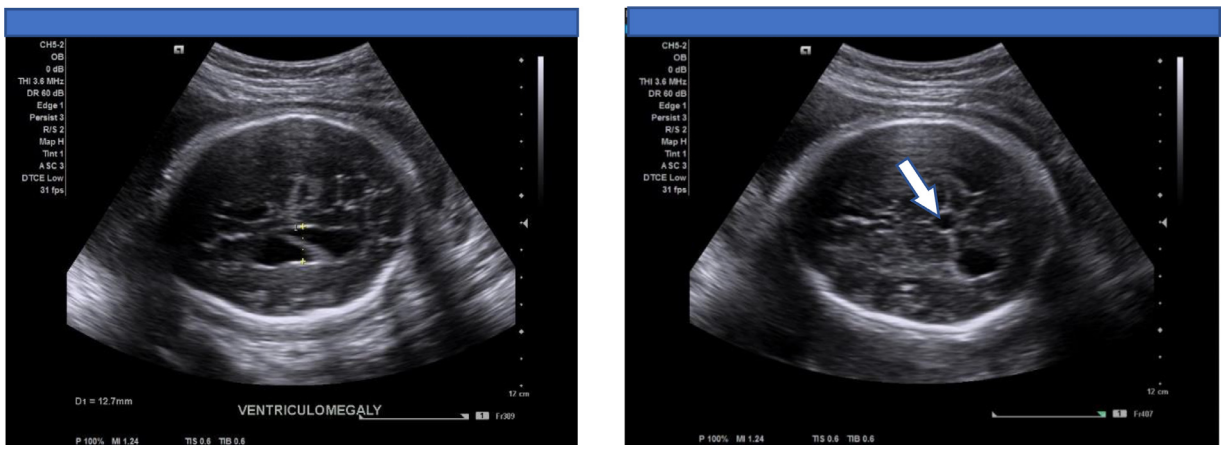

Fig. 3 – Sonogram of the fetal head (axial view) shows dilatation of the lateral ventricle (measuring 12.7 mm) and dilated third ventricle (white arrow).

Upon delivery, an open spinal defect and associated lesion with visible neural tissues at the lumbosacral region was observed in the demised fetus.

A postdelivery pelvic ultrasound scan confirmed no retained products of conception. The patient was further discharged with prescribed pain medications and antibiotics (Ciprofloxacin 500 mg and Metronidazole 500 mg). Immediate follow-up care included monitoring for any immediate post-procedure complications and providing psychological support as needed. The patient did not report any complications after her treatment and discharge.

Discussion

Spina bifida, classified as either open or closed; is a congenital neural tube defect occurring in approximately 1 per 1,000 live births [1].

Myelomeningocele, the most severe and clinically significant form of open spina bifida is characterized by herniation of the spinal cord and its contents through a bony defect in the posterior elements of the spine [1,2].

The cause of myelomeningocele is multifactorial in nature. Maternal factors such as malnutrition, low or no folate supplementation, alcohol consumption, smoking, diabetes and obesity have been linked to myelomeningocele [2]. Also, exposure to radiation, teratogenic agents and pollutants such as pesticides and organic solvents have been identified as environmental risk factors for myelomeningocele. Furthermore, genetic factors and the presence of other chromosomal anomalies such as trisomy 18 or 13 are predisposing factors for the development of myelomeningocele [1,5].

The spinal cord during its development in the 2nd to 6th week of embryogenesis involves 3 phases namely gastrulation, primary neurulation and secondary neurulation. During neurulation, formed neural folds are fused together to form the neural tube. The cranial and caudal ends of the neural tube are closed by the end of the 4th week and this completes the primary neurulation. Failure of closure of the caudal aspect of the neural tube at this stage often times leads to exposed neural tissues and meningeal lining, leading to the

formation myelomeningocele as the pregnancy progresses [2].

The primary mechanism leading to Chiari II malformation, is with the chronic leakage of cerebrospinal fluid (CSF) through the open neural tube defect in myelomeningocele. This leakage leads to decreased intracranial pressure and subsequent reduction in the posterior fossa volume. This causes a pressure gradient across the foramen magnum and as a compensatory mechanism, the cerebellar vermis, tonsils, and brainstem herniate downward into the spinal canal to occupy the reduced space, leading to the characteristic hindbrain herniation seen in Chiari II malformation. This abnormal downward displacement can lead to several secondary effects, including obstruction of the CSF pathways at the craniovertebral junction, leading to hydrocephalus (excessive accumulation of CSF within the brain ventricles) [4,9].

Ultrasonography and magnetic resonance imaging (MRI) have been widely utilized as gold standard diagnostic tools for myelomeningocele and Chiari II malformation both in the antenatal and neonatal period [10,11]. Although certain embryonic features in first trimester ultrasound has been described as useful for the early detection of open spina bifida [12–14], it is worth noting that visibility of obvious features and the experience of the ultrasound practitioner are factors that can influence the identification of these anomalies with certainty [15]. This is relatable to our case report; whereby a previous ultrasound performed in the first trimester in another health facility could not detect spina bifida. This might have influenced the patient's decision for not regularly attending antenatal care as she may had assumed she had a normal pregnancy.

Recent studies have identified the third trimester ultrasound to be sensitive for the first-time detection of certain fetal anomalies such as spinal defects, intracranial pathologies, obstructive bowel and genitourinary anomalies [16–19]. Most detected gross anomalies in the third trimester are those that might have been missed in earlier scans in the first and second trimester, as well as anomalies that develop and become visible as the pregnancy progresses [17]. Undetected fetal anomalies in the first and second trimester can also be attributed to lack of adequate periconceptional care. This is relatable to this current case report where the patient had poor antenatal visit history and had never taken any periconceptional folate supplementation. This could have contributed to her fetus developing meningomyelocele since daily periconceptional folate supplementation has been seen to be effective in preventing neural tube defects [20–22].

As myelomeningocele and associated Chiari II malformation poses significant neurological impairment and life-threating conditions such as respiratory deficit to the neonate, management of this anomaly involves a multidisciplinary team and counselling sessions to the expectant mother and family. Medical management include medical termination of pregnancy if significant deficit and poor prognosis are envisioned. Surgical repair (either prenatal or postnatal) is also an option for the management of myelomeningocele and associated Chiari II malformation. Prenatal (fetal) surgical repair is however mostly advocated for since it has been seen to be more effective in the preservation of neurological function thereby improving outcomes [6–8].

For our case report, the patient opted for medical termination after receiving adequate counselling from her attending obstetrician.

Conclusion

Adequate periconceptional care, coupled with ultrasound screening for fetal anomalies at every stage of the pregnancy remains crucial for the detection and management of neural tube and intracranial anomalies. This case report therefore highlights the need for clinicians to request for detailed ultrasound scanning in women who present late for antenatal visit (in the third trimester); and those women with poor folate supplementation. This will allow for the detection of anomalies that may have been missed earlier on in the course of their pregnancies.

Patient consent

Written informed consent for the publication of this case report was obtained from the patient.

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