



## Spina Bifida Aperta Ultrasound and Histopathology Examination: About a Case Report

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### Abstract

Spina bifida aperta is a congenital malformation characterized by incomplete closure of the neural tube during fetal development. Prenatal diagnosis of spina bifida aperta plays a crucial role in enabling early interventions and improving patient outcomes. This case report aims to present the importance of first-trimester ultrasound and histopathology examination in diagnosing spina bifida aperta.

A pregnant woman at 13 weeks gestation presented for routine prenatal care. Transabdominal ultrasound examination revealed an abnormality consistent with a neural tube defect. Detailed assessment of the fetal spine using high-frequency transducers and three-dimensional ultrasound showed a midline defect with a sac-like protrusion containing cerebrospinal fluid, indicative of spina bifida aperta. Subsequent magnetic resonance imaging confirmed the ultrasound findings.

Early detection of spina bifida aperta during the first trimester is essential for appropriate management and counseling. Ultrasound imaging plays a pivotal role in the antenatal diagnosis of spina bifida aperta. It provides detailed visualization of the neural tube and associated structural abnormalities. In complex cases, magnetic resonance imaging can offer further anatomical delineation.

Histopathological examination remains a valuable adjunct to prenatal diagnosis, providing a definitive confirmation of spina bifida aperta since gross macroscopic examination aids in assessing the extent and nature of the defect, while microscopic analysis offers insights into the histological changes associated with the condition.

### Introduction

Spina bifida aperta is a severe congenital malformation characterized by the incomplete closure of the neural tube during embryonic development. It is the most common Central Nervous System (CNS) malformation compatible with life, affecting approximately 1 to 2 per 1,000 live births worldwide, represented by the lack of dorsal closure of the vertebrae which lack neural arches and may be located at any level along the spine. These defects can be classified into closed when the spinal defect is skin covered (spina bifida occulta) or open (spina bifida aperta) when the lesion site is not covered by skin and involves abnormalities of the meninges and/or the spinal cord (meningocele/myelomeningocele).

This condition results in significant morbidity and lifelong disability, making early diagnosis and intervention crucial for optimizing outcomes.

Prenatal detection of spina bifida aperta has evolved significantly over the years, with advancements in imaging techniques allowing for accurate and timely diagnosis. Ultrasonography, particularly during the first trimester, has emerged as a valuable tool in the antenatal evaluation of fetal anomalies. High-resolution transabdominal and transvaginal ultrasound approaches enable detailed visualization of the developing neural tube, facilitating the identification of structural abnormalities associated with spina bifida aperta.

In recent years, these approaches have been increasingly utilized in first-trimester ultrasound examinations, offering the possibility of early intervention and improved outcomes. Additionally, histopathological examination of terminated pregnancies provides valuable insights into the underlying pathology and helps confirm the ultrasound findings. This integrated approach allows a better assessment of the condition, aiding in counseling and appropriate management strategies.

This case report aims to present a detailed analysis of a specific case of spina bifida aperta

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diagnosed during the first trimester in order to contribute to the existing body of knowledge on this serious condition, particularly in terms of its prenatal diagnosis and associated histopathological features.

The combined use of these diagnostic modalities provides a comprehensive understanding of the condition, emphasizing the importance of early detection and accurate characterization for effective clinical decision-making.

Furthermore, this report underscores the significance of multidisciplinary collaboration between obstetricians, radiologists, and pathologists in the evaluation and management of fetal anomalies. Ultimately, improved diagnostic capabilities of spina bifida aperta can lead to enhanced prenatal counseling and better outcomes for affected pregnancies.

## Case Presentation

A 27-year-old primigravida woman presented for routine prenatal care at 13 weeks of gestation. The patient had no significant family history related to neural tube defects. Transabdominal ultrasound screening was performed using a high-resolution transducer and three-dimensional ultrasound.

The ultrasound examination revealed a midline defect in the lumbosacral region of the fetus's spine. A detailed assessment of the defect showed a sac-like protrusion containing cerebrospinal fluid, indicating the presence of spina bifida aperta. The visualization of the defect and the associated findings were consistent with the diagnosis of this neural tube defect.

To further confirm the diagnosis and obtain additional anatomical details, Magnetic Resonance Imaging (MRI) was conducted. The MRI findings were in concordance with the ultrasound results, confirming the presence of the spinal defect and providing precise delineation of the associated structural abnormalities.

Given the diagnostic findings and after thorough counseling, the patient opted for the termination of the pregnancy. Subsequently, a histopathological examination was performed on the fetus.

Gross examination of the fetus revealed an open defect in the spinal column (Figure 2), confirming the presence of spina bifida aperta. The defect corresponded to the previously identified midline defect observed during the ultrasound and MRI examinations.

Microscopic analysis of the spinal cord tissue obtained from the affected area demonstrated dysplastic changes, indicative of abnormal development. The examination also revealed the herniation of neural elements through the defect, further confirming the diagnosis of spina bifida aperta.

Histological staining techniques allowed for detailed evaluation of the cellular architecture and structural changes within the spinal cord tissue. The dysplastic features included disorganized cell layers, abnormal neuronal migration, and disrupted neural connectivity. These histopathological findings provided further insight into the underlying pathology associated with spina bifida aperta.

The combination of the prenatal ultrasound screening, MRI, and histopathological examination provided a better understanding of the diagnosis of spina bifida aperta in this case. The initial ultrasound findings were subsequently validated by the MRI, and the histopathological examination served as definitive confirmation, providing detailed insights into the structural and cellular abnormalities originally causing the condition.

## Discussion

Open spina bifida, also known as spina bifida aperta is a neural tube defect involving the lack of closure of vertebral arches and associated meninges and/or spinal cord abnormalities. Ultrasound examination being the gold standard for the diagnosis of spina bifida aperta, represents the main imaging tool used to ascertain this diagnosis early in gestation.

While three-dimensional ultrasound is necessary to detect the level and the size of the defect, Magnetic Resonance Imaging (MRI) represents a more sensitive tool, giving specific information of the defect and associated anomalies, playing an important role in ruling out differential diagnosis. Due to the advent of MRI use, it is possible

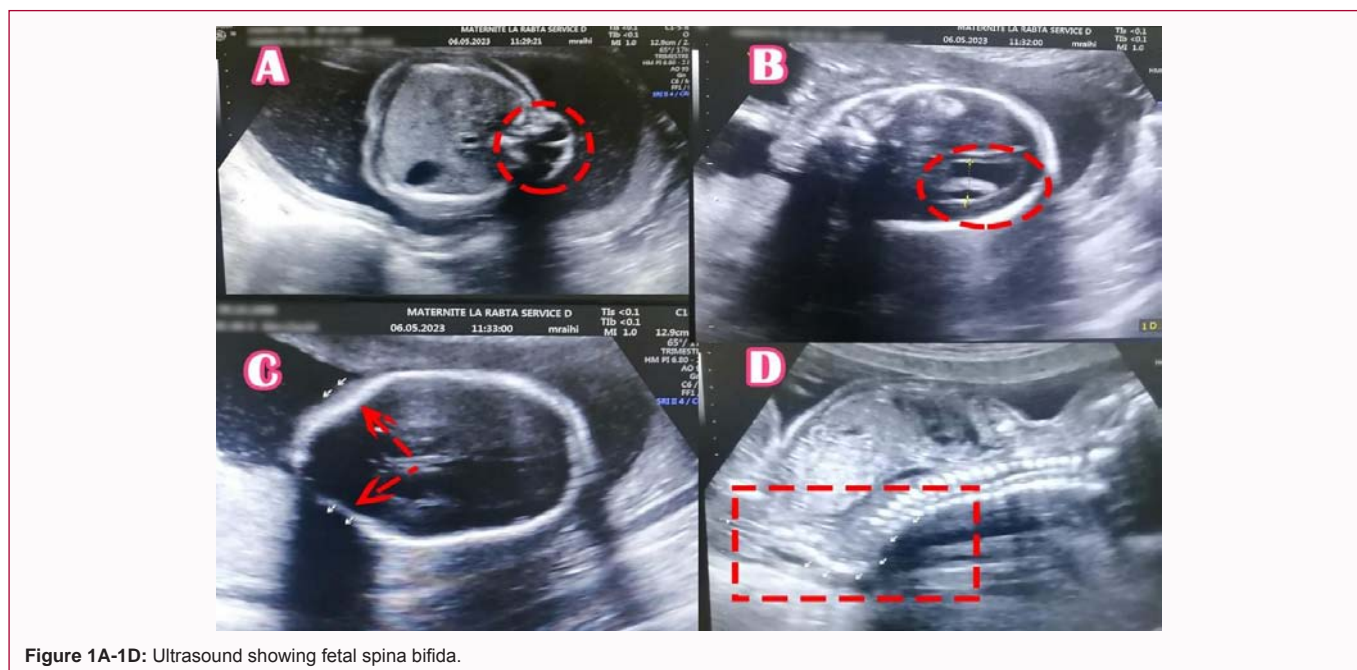


Figure 1A-1D: Ultrasound showing fetal spina bifida.



**Figure 2:** Histological characterization of spina bifida aperta.

today to achieve in utero treatment of fetuses with this pathology.

The case presented here highlights the significance of utilizing first-trimester ultrasound screening and subsequent histopathological examination in diagnosing spina bifida aperta [1]. This integrated approach involving imaging and histology provides more accurate diagnosis, counseling, and management decisions [2].

First-trimester ultrasound screening plays a crucial role in the prenatal diagnosis of spina bifida aperta. High-resolution transabdominal and transvaginal ultrasound techniques allow for detailed visualization of the fetal spine and identification of structural abnormalities [3,4].

In this case, multiple findings consistent with the characteristic features of spina bifida aperta (a midline defect in the lumbosacral region, accompanied by a sac-like protrusion containing cerebrospinal fluid) were detected early in gestation using a well oriented, performed and accurate first trimester ultrasound screening allowing early intervention and appropriate management planning [5].

To validate the ultrasound findings and obtain additional anatomical details, MRI was performed confirming the presence of the spinal defect and providing precise delineation of the associated structural abnormalities [3]. This advanced imaging tool is particularly useful in complex cases where detailed visualization and characterization of the defect are required. The integration of ultrasound and MRI imaging enhances diagnostic accuracy and aids in surgical planning if required [6].

Histopathological examination serves as a definitive confirmation of the prenatal diagnosis of spina bifida aperta. In this case, following the termination of the pregnancy, a thorough examination of the fetus was conducted [7]. Gross macroscopic examination revealed an open defect in the spinal column (Figure 2), corroborating the ultrasound and MRI findings. The size and location of the defect were carefully documented, providing important information for clinical management [2].

Microscopic analysis of the spinal cord tissue obtained from the affected area further supported the diagnosis of spina bifida

aperta. Dysplastic changes, characterized by disorganized cell layers, abnormal neuronal migration, and disrupted neural connectivity, were observed [8]. These histological findings provide valuable insights into the underlying pathological mechanisms involved in spina bifida aperta and therefore help understanding the histopathological features of the condition contributing to a better understanding of its etiology and pathogenesis [9].

The integrated use of first-trimester ultrasound screening and histopathological examination allows for a multidimensional evaluation of spina bifida aperta. Early detection of the condition during pregnancy enables appropriate counseling and management strategies [10]. It provides an opportunity for parents to make informed decisions regarding the continuation of the pregnancy and prepares healthcare professionals for any potential surgical interventions or postnatal care requirements [11].

Moreover, the combined expertise and collaborative input of obstetricians, radiologists, and pathologists is crucial in the complete evaluation and management of spina bifida aperta thus these specialties ensure accurate diagnosis, facilitate multidisciplinary discussions, and optimize patient care.

This case report emphasizes the importance of integrating efficient first-trimester ultrasound screening and histopathological examination in the diagnosis of spina bifida aperta [11]. This comprehensive approach enhances prenatal care and contributes to improved outcomes for patients affected by spina bifida aperta. Further research and advancements in diagnostic techniques will continue to improve our understanding and management of this complex congenital condition.

## Conclusion

In conclusion, the presented case report highlights the significance of combining first-trimester ultrasound screening and histopathological examination in diagnosing spina bifida aperta allowing more informed decision-making, and optimized patient care.

Indeed, early detection during pregnancy enables timely interventions and counseling.

Continued research and advancements in diagnostic techniques will further enhance our understanding and improve outcomes for individuals affected by spina bifida aperta.

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