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Prenatal diagnosis of lipomyelomeningocele by ultrasound and magnetic resonance imaging (MRI)

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Summary

Objective: The authors report a case of a lipomyelomeningocele with tethered cord, revealed on prenatal ultrasonography and confirmed by fetal magnetic resonance imaging (MRI). *Materials and Methods:* A 32-year-old woman, gravida 1 para 1 underwent the routine second trimester prenatal ultrasound scan at 22^{+5} weeks of gestation at the present hospital. *Results:* The scan indicated an echoic semisolid subcutaneous mass covered by skin, posterior to the lumbosacral spinal canal of the fetus. Based on the findings indicating occult dysraphism, a fetal MRI examination was conducted, revealing that the mass was extending to the spinal cord, tethering the cauda equina. The diagnosis of lipomyelomeningocele was established. *Conclusion:* Lipomyelomeningocele is a form of closed neural tube defect with unclear predisposing factors. Its prevalence ranges between 0.3 and 0.6 per 10,000 live births. It leads to progressive conus tethering with associated neurological, urinary, and gastrointestinal deficits, demonstrating the importance of prenatal diagnosis.

Key words: Lipomyelomeningocele; Spinal dysraphism; Occult spina bifida; Tethered cord; Fetal MRI; Prenatal ultrasound.

Introduction

Spinal dysraphism is a medical term referring to a broad-spectrum group of congenital defects resulting from incomplete neural tube closure during the early stages of the developing fetus. The cause is multifactorial, involving both genetic and environmental factors [1]. These malformations can be categorized into open and occult forms [2]. The occult forms have normal skin cover in contrast with open ones that are associated with skin defects [2, 3]. Their clinical presentation varies from asymptomatic to causing serious disabilities to the patient with lethal complications [4].

Embryologically, the expansion of the neural tube and subsequent closure is completed by day 28. Open defects occur when the caudal neuropore fails to close [2]. The secondary neurulation sets the spinal cord formation [2]. Several types of closed spinal dysraphisms result from embryological abnormalities during this phase [2, 5]. Those that arise from premature disjunction result in fusion of the spinal cord with fatty elements, the most common of which is a lipomyelomeningocele [5].

Lipomyelomeningocele is a rare but complicated defect, lying in the spectrum of occult neural tube defects. It is actually a form of occult spinal dysraphism in which a subcutaneous fibrofatty mass traverses the lumbodorsal fascia, causes a spinal laminar defect, displaces the dura, and infiltrates and tethers the spinal cord [6]. The prevalence of lipomyelomeningocele ranges between 0.3 and 0.6 per 10,000 live births [5,7]. The general prevalence of spinal dysraphism has declined internationally in the last few decades due to the better nutrition of women, folic acid supplementation, and establishment of ultrasound scan and biochemical markers as routine prenatal screening [2, 5, 8]. However, rates of lipomyelomeningocele do not seem to have been reduced according to recent studies, suggesting that the pathogenesis of lipomyelomeningocele is fundamentally different from that of other neural tube defects [7, 9]. According to the modified Chapman classification, that takes into consideration the anatomical relation of the lipoma with the cord interface, lipomyelomeningocele is subdivided into dorsal, transitional, and caudal [10-12]. As the subcutaneous lipoma infiltrates the spinal cord through the defect in the lumbodorsal fascia, the upward movement of the conus medullaris during axial growth may be limited and thus may lead to progressive conus tethering with associated neurological, urinary, and gastrointestinal deficits- the sequelae of a tethered cord [13]. Bowel and bladder function usually deteriorate prior to motor function or sensation [14]. However 62.5% of patients with lipomyelomeningocele have been found to be neurologically asymptomatic prior to six months of age, while only 29.3% are asymptomatic after six months of age [15]. No affected children remain asymptomatic after the age of five years [16]. The disease progression can result in frequent urinary tract infections and neurogenic bladder and bowel incontinence or constipation, as well as leg length discrepancy, foot deformities, gait abnormalities, scoliosis, spasticity, and back and leg pain [5, 14, 15]. Earlier intervention to correct a tethered cord increases the chance that its sequelae will be reversible [17]. These facts demonstrate the

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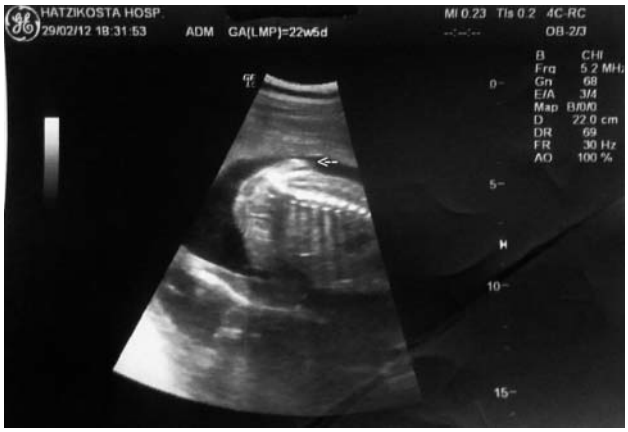

Figure 1. — The ultrasound revealed a subcutaneous mass covered by skin, protruding posterior to the lumbosacral spinal canal of the fetus (arrow head).

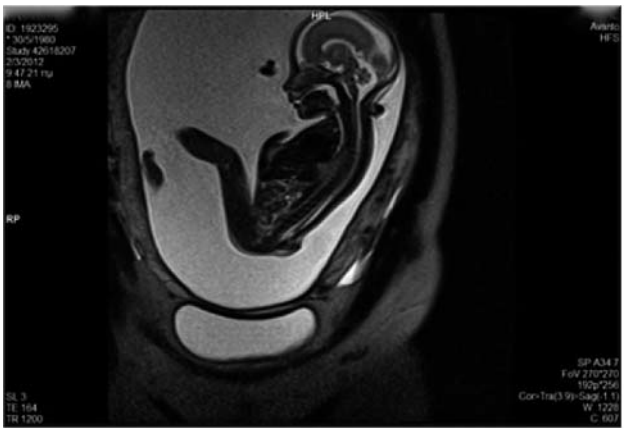

Figure 2. — A more detailed image of the lesion by MRI. The lipoma extends to the spinal cord, tethering the cauda equina.

importance of prenatal diagnosis of this rare medical entity, so that the parents decide after proper consultation on the preservation or termination of the current gestation and so that subsequently timely postnatal surgical intervention for the optimal treatment of the neonate is achieved. The authors describe a case of a lipomyelomeningocele with tethered cord, which was indicated on prenatal ultrasonography and confirmed by fetal magnetic resonance imaging (MRI).

Materials and Methods

A 32-year-old woman, gravida 1 para 1 underwent the routine second trimester prenatal ultrasound scan at 22^{+5} weeks of gestation at the Obstetrics' Department of the present Hospital. The first trimester scan was normal with 1.7 mm measured nuchal translucency of a female fetus, the prenatal screen was unremarkable, and her pregnancy had been uneventful up to that point. Her medical and obstetric record was free and she had no family history of neural tube defects or other congenital malformations and no history of exposure to known teratogens. She had been on five mg of folic acid since the 5^{th} week of gestation.

Results

The two-dimensional (2D) ultrasound examination that was performed showed normal amniotic fluid index (AFI) and single umbilical artery was noted. No cranial ultrasound abnormalities were identified- the posterior fossa and ventricles were normal, excluding anomalies like ventricular dilatation or Arnold-Chiari malformation. An echoic semisolid subcutaneous mass covered by skin was observed to protrude posterior to the lumbosacral spinal canal of the fetus (Figure 1). The color Doppler image revealed no blood flow in the lesion. The remaining fetal anatomy appeared normal. Maternal serum alpha-fetoprotein (AFP) level was not obtained. Based on the findings leading to a diagnosis of occult dysraphism, an ultrafast fetal MRI examination was conducted. The previously described lesion measured 24.5 x.10.8 x 14.5 mm. The additional information added by this examination was that the spinal cord was lying low and the subcutaneous mass was extending to the spinal cord, tethering the cauda equina (Figure 2). The diagnosis of lipomyelomeningocele was established. The parents consulted neurosurgeons and decided to terminate the pregnancy. The next gestation of the couple resulted in the birth of a healthy neonate with no neural tube defect.

Discussion

Lipomyelomeningocele is a form of closed neural tube defect with unclear predisposing factors [5]. Due to the risk of deteriorating neurological and urological function secondary to a tethered spinal cord, it is crucial to identify this condition prenatally for timely intervention. Familial forms of lipomyelomeningocele are rare [18, 19] but parents should be aware that these lesions could occur in subsequent pregnancies and thus that they should undergo early and detailed prenatal ultrasound examination in the next gestation.

The authors describe the prenatal diagnosis of a caudal-type lipomyelomeningocele. There are not many similar published case reports in the literature. It is important for the obstetrician to bear in mind that while most spinal dysraphisms are associated with abnormal ultrasound findings of the fetal brain, only 3.6% of the lipomyelomeningocele cases present with brain malformations [20]. As a result, in order to detect these occult anomalies, it is of great importance to completely scan the fetal spine, especially in the lumbosacral region, along with the fetal head [21]. MRI is indicated as the second-line prenatal diagnostic tool for a more detailed examination of the fetal anatomy when ultrasound indications for malformations of the central nerv-

ous system exist [22]. Its effectiveness in providing more accurate information is already well known. The Safety Committee of the Society for MRI has concluded that prenatal MRI is indicated when other non-ionizing methods are inadequate or when the MRI examinations will provide critical information that would otherwise require the use of ionizing radiation [23].

Conclusion

The existence of lipomyeloeningocele should be included in the differential diagnosis of cases of prenatally revealed sacral lesions accompanied with spinal dysraphism. Careful prenatal imaging may lead to the establishment of a prenatal diagnosis of this entity and contributes to discrimination between fetuses with malformations with a favorable prognosis from those with a poor one. As a result, parents are offered more accurate prenatal counseling from specialists regarding the clinical course and possible future disabilities of the offspring so that they can decide to preserve or terminate the current pregnancy, bearing in mind the possibility of recurrence in a subsequent pregnancy.

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