Case report

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Magnetic resonance imaging diagnosis of an open spina bifida defect

Milan Stefanović^{1,2}, Vesna Stokanović³, Predrag Vukomanović^{1,2}, Vanja Dimitrov², Aleksa Stefanović²

¹Univerzitet u Nišu, Medicinski fakultet, Niš, Srbija

²Univeristy Clinical Center Niš, Clinic of Gynecology and Obstetrics, Niš, Serbia

³Univeristy Clinical Center Niš, Radiology Center, Niš, Serbia

Contact: Milan Stefanović

81 dr Zoran Djindjić Blvd., 18000 Niš, Serbia

E-mail: milstef64@gmail.com

Spinal neural tube defects are a group of disorders that allude to spina bifida, anencephaly, interspinal lipoma, congenital dermal sinus, simple tethered cord and other diseases that can cause neurological problems such as motor and/or sensory disorders and urinary and/or fecal incontinence. Usually, they can be diagnosed with a combined interpretation of alpha fetoprotein and ultrasound imaging. However, sometimes the ultrasound image quality is poor and necessitates the use of a higher resolution imaging technique – MRI.

A woman in her 18th week of pregnancy was scheduled for a routine prenatal check-up. Upon examination, a mild, yet, suspicious lemon sign of the fetus's cranium had been observed. The sagittal view of the fetus's spine was normal, however the transversal view of the spine could not be assessed. The complete ultrasound examination was of lower quality due to fetal and maternal factors. Thus, a decision for a fetal MRI had been made. The MRI scan confirmed an open spina bifida.

Since MRI scanning is a greater tool than ultrasound, whenever it isn't possible to conclusively diagnose a neural tube defect via ultrasound for any reason, an obstetrician should opt for an MRI scan to confirm the condition.

Keywords: magnetic resonance imaging, neural tube, defect, diagnosis

Prikaz slučaja

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Uspostavljanje dijagnoze otvorenog defekta neuralne cevi pomoću magnetne rezonance

Milan Stefanović^{1,2}, Vesna Stokanović³, Predrag Vukomanović^{1,2}, Vanja Dimitrov², Aleksa Stefanović²

¹Univerzitet u Nišu, Medicinski fakultet, Niš, Srbija

²Univerzitetski klinički centar Niš, Klinika za ginekologiju i akušerstvo, Niš, Srbija

³Univerzitetski klinički centar Niš, Centar za radiologiju, Niš, Srbija

Kontakt: Milan Stefanović

Bulevar dr Zorana Điniđića 81, 18000 Niš, Srbija

E-mail: milstef64@gmail.com

Defekti neurvne cevi su grupa poremećaja koja uključuju spinu bifidu, anencefaliju, interspinalni lipom, urođeni dermalni sinus, jednostavnu vezanu vrpcu i druge bolesti koje mogu izazvati neurološke probleme kao što su motorni i/ili senzorni poremećaji i urinarna i/ili fekalna inkontinencija. Obično se mogu dijagnostikovati kombinovanom interpretacijom alfa fetoproteina i ultrazvučnog snimanja. Međutim, ponekad je kvalitet ultrazvučne slike loš i zahteva upotrebu tehnike snimanja veće rezolucije – magnetnu rezonancu.

Žena u 18. nedelji trudnoće zakazana je za rutinski prenatalni pregled. Nakon pregleda, uočen je blag, ali sumnjiv znak limuna na lobanji fetusa. Sagitalni pogled na kičmu fetusa je bio normalan, ali transverzalni presek kičme nije mogao da se adekvatno sagleda. Kompletan ultrazvučni pregled je bio lošijeg kvaliteta zbog fetalnih i majčinskih faktora. Zbog toga je doneta odluka za MR fetusa. MR skeniranje je potvrdilo otvorenu spinu bifidu.

Pošto je MRI skeniranje bolji alat od ultrazvuka, kad god nije moguće konačno dijagnostikovati defekt nervne cevi ultrazvukom iz bilo kog razloga, akušer bi trebalo da se odluči za MRI skeniranje kako bi se potvrdilo stanje.

Ključne reči: magnetna rezonanca, neuralna cev, defekt, dijagnoza

Introduction

Spinal neural tube defects are a group of disorders that allude to spina bifida, anencephaly, interspinal lipoma, congenital dermal sinus, simple tethered cord and other diseases that can cause neurological problems such as motor and/or sensory disorders and urinary and/or fecal incontinence. (1-3)

The incidence rate of spinal neural tube defects is 0.5-2%, and the incidence of spina bifida is around 3,63 % in the US and 18.6% worldwide. (4,5)

Spina bifida itself is a condition where the spinal column fails to close and remains split-bifid. This is a result of a neural tube failing to close during the fourth week of embryonic development.

Spina bifida can be presented as a closed defect - *occulta*, or a more common and more severe open defect – *aperta*. (6,7)

People born with spina bifida have lifelong consequences. (8)

This makes early diagnosis and prevention crucial in reducing morbidity and for optimizing outcomes.

In recent times ultrasonography combined with alpha fetoprotein analyses is integral to prenatal diagnosis of spina bifida. (9,10)

However, AFP can be elevated in other conditions in pregnant women such as gastroschisis, omphalocele, osteogenesis imperfecta etc. (11) Ultrasound also has its shortcomings. Low resolution, factors that can affect image quality or visibility of structures. (12,13)

In cases where ultrasound findings aren't conclusive, a physician can opt for a higher quality imaging method- an MRI.

Since its beginnings MRI (Magnetic Resonance Imaging) has evolved into a highly sophisticated imaging method particularly suitable for visualizing soft tissues. Modern MRIs have reduced artifacts caused by fetal movements, which makes them reliable in visualizing in utero disorders. (14)

Case

A pregnant woman at 18 weeks of gestation was scheduled for routine prenatal care.

Ultrasound examination showed an inconclusive lemon sign. The posterior cranial fossa had not been accessible for visualization.

The sagittal view of the spine seemed to be normal, but the transversal view could not be performed.

Transabdominal ultrasound findings were indicative for a neural tube defect. However, the tissue resolutions, maternal obesity, fetal malposition, an anterior placenta, along with the inability to view the spine transversally, caused the impossibility of definitive conclusion.

Thus, a decision had been made for the patient to undergo magnetic resonance imaging in order to confirm the ultrasound findings.

The MRI findings are presented in **Figure 1.**.

Figure 1. a) Sagittal T2W SSFSE tomogram. Posterior fossa is small. Cerebellar tonsils are pointed, compressed, protruding through foramen magnum (arrow). IV ventricle is compressed. Cisterna magna is effaced. Discontinuity of the skin and subcutaneous tissue on a long segment involving lower thoracic, complete lumbar and part of the sacral region is seen (dotted arrow). b) Axial T2W SSFSE tomogram just below the level of foramen magnum. Cerebellar tonsils are wrapped around medulla, filling the foramen magnum. c) Axial T2W SSFSE tomogram through endocranium at the level of lateral ventricles. Lateral ventricles are on the upper limit of the normal size, with maximum width of 10mm. d) Axial T2W SSFSE tomogram through abdomen below the level of kidneys. Posterior vertebral elements are lacking. The neural tissue forming the placode is at the level of the adjacent skin surface and exposed to the

The MRI images were interpreted by a radiologist and concluded that the scans were positive for neural tube defect – specifically spina bifida aperta.

Afterwards, on an ethical committee meeting, a decision had been made to approve an elective abortion due to medical reasons.

The medical abortion had been performed and the extracted fetus had an open neural tube defect – spina bifida aperta (**Figure 2**).



Figure 2. The fetus with an apparent neural tube defect – open spina bifida

Discussion

Spina bifida is not an uncommon birth defect and it carries with itself potentially lifelong consequences. (8)

Because of this, prenatal diagnosis and adequate assessment are needed to optimize the outcomes.

The optimal period for ultrasound scanning of neural tube defects is 11-14 weeks of gestation. (9)

Even though ultrasound has come a long way, it is by no means a perfect diagnostic tool.

The problems of ultrasound imaging such as tissue resolutions, maternal obesity, fetal malposition, an anterior placenta, late pregnancy, olygohydramnion, abdominal scar in pregnant women can all lead to poor image quality and inability to adequately diagnose spina bifida. (12,13)

This makes MRI imaging the next step in diagnostics since it is a superior visualization method compared to ultrasound. (15)

Conclusion

Since MRI scanning is a greater tool than ultrasound, whenever it isn't possible to conclusively diagnose a neural tube defect via ultrasound for any reason, an obstetrician should opt for an MRI scan to confirm the condition.

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