NEURAL TUBE DEFECTS: A CASE OF LUMBAR MYELOMENINGOCELE

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1- Introduction/objectives

Neural tube defects (NTDs) are relatively common congenital anomalies: studies have reported a prevalence of 9 per 10,000 births in Europe; they develop normally during the fifth and sixth weeks of gestation and they may be open or closed. Open NTDs comprise 80% of NTDs, most of which are myelomeningoceles[1].

Myelomeningocele is a devastating congenital defect of the Central Nervous System (CNS), which is characterized by protrusion of the meninges and spinal cord through open vertebral arches that could lead to lifelong paralysis[2]. Besides, it can cause varying degrees of neurologic impairment, depending upon the level of the lesion, and can impair cognitive function, depending upon the presence and degree of associated ventriculomegaly or associated genetic syndrome.

We present a case of lumbar myelomeningocele to demonstrate the importance of prenatal diagnosis and ensure a better postnatal management.

2- Methods

A descriptive case report.

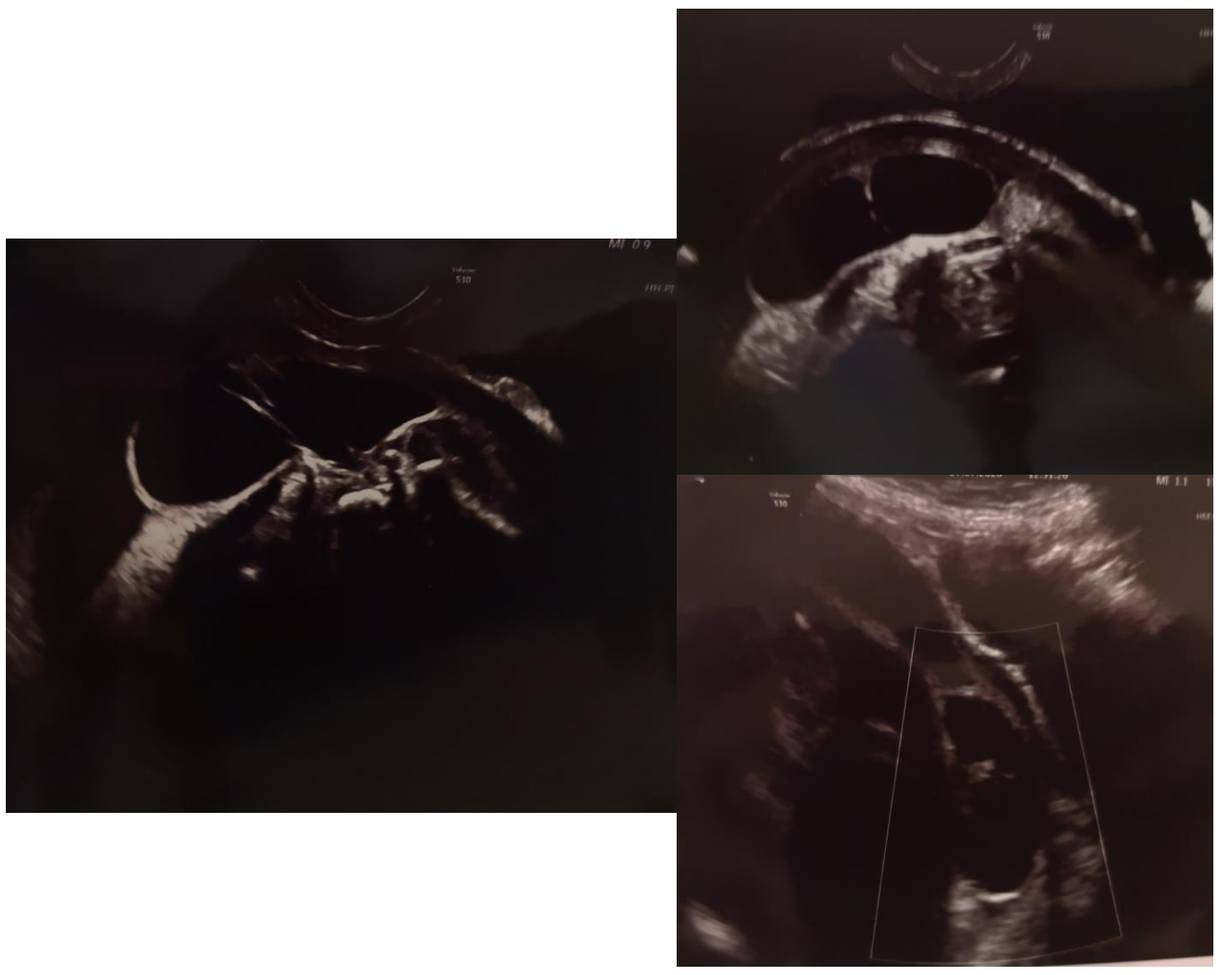
3- Results

A 33-year-old woman, gravida 1, was referred to our centre at 25 weeks of gestation after diagnosis of gestational diabetes in the second trimester. The patient did not have any relevant medical or surgical history and there was no family history of birth defects. Until then, she has had a normal gestational course with low risk of aneuploidies in the first trimester screening and normal fetal scans, both in the 1srt and 2nd trimesters.

Her gestational diabetes remained well controlled with only non-pharmacological measures. The sonographic examination performed at 29 weeks of gestation revealed a cystic exophytic mass with septations of 33 x 35 mm arising from the terminal portion of the lumbar spine, probably related to myelomeningocele. No other abnormalities were detected in this scan. Magnetic resonance imaging (MRI) was performed, which confirmed the diagnosis.

The patient was referred to a centre with pediatric surgery support. Follow-up ultrasounds were performed and showed no increase in the size of the mass.

The patient had an elective caesarean section scheduled for 39 weeks; however, she was admitted at the emergency department of our hospital at 37 weeks and 6 days due to premature rupture of membranes. We then transferred the patient to a central hospital with pediatric surgery support.



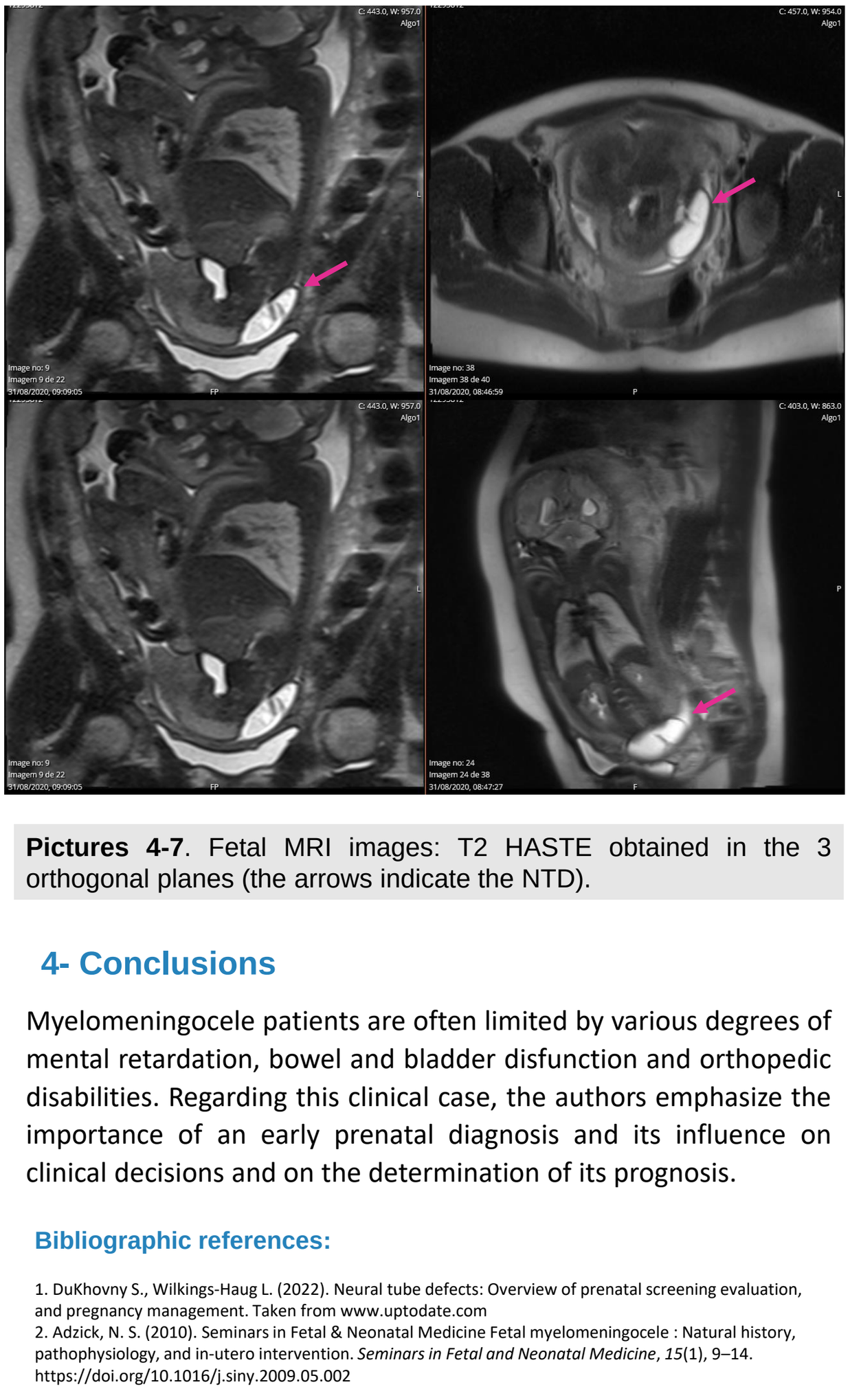
Pictures 1-3. Ultrasound images of the myelomeningocele obtained via transabdominal and transvaginal routes.

A male baby weighing 3100g was delivered by caesarean section. At birth, the lumbar myelomeningocele was confirmed. The new-born was admitted to the NICU, having undergone primary closure of the NTD on the 3rd day of life, without intercurrences.

At 12 days of life, a ventriculo-peritoneal shunt was required due to worsening hydrocephalus. The baby was discharged 44 days after birth, with a good postoperative evolution.

Currently, the 12-month-old child has sphincter incontinence and can mobilize all four limbs, although it does not yet have autonomous gait.

He maintains follow-up in pediatrics, neurosurgery, orthopedics, pediatric surgery and physiatrics consultations.



Pictures 4-7. Fetal MRI images: T2 HASTE obtained in the 3 orthogonal planes (the arrows indicate the NTD).

4- Conclusions

Myelomeningocele patients are often limited by various degrees of mental retardation, bowel and bladder disfunction and orthopedic disabilities. Regarding this clinical case, the authors emphasize the importance of an early prenatal diagnosis and its influence on clinical decisions and on the determination of its prognosis.

Bibliographic references:

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