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LIPOMYELOMENINGOCELE: ANTENATAL DIAGNOSIS OF CLOSED SPINA BIFIDA

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BACKGROUND

Spina bifida develops during closure of the neural tube. Open spina bifida is characterised by herniation of exposed neural tissue through a cleft in the spinal column, unlike closed spina bifida in which neural tissue is protected by intact overlying skin. Lipomyelomeningocele is a type of closed spina bifida, with an estimated prevalence of 2.5 per 10 000 live births. A subcutaneous lipoma extends through the lumbodorsal fascia and dura, resulting in spinal cord tethering. Postnatal clinical symptoms may include lower limb motor and sensory changes, neurogenic bladder, spinal scoliosis or leg length discrepancy.

CASE

A 42-year-old multiparous woman was diagnosed at morphology ultrasound with a fetal closed neural tube defect; there was no family history of spina bifida. She had a low risk NIPT and normal 12-week ultrasound.

Morphology ultrasound demonstrated an avascular soft tissue mass (22mm x 17mm x 18mm) over the sacrum with splaying of vertebrae at S2/S3 and an echogenic mass arising from the spinal canal extending to the sacral mass with ultrasound evidence of tethering. Both the intracranial and lower limb anatomy appeared within normal limits.

The diagnosis of lipomyelomeningocele was also confirmed on fetal MRI. The woman received counselling at a dedicated spina bifida clinic and decided to proceed with termination of the pregnancy. Post-mortem was consistent with lipomyelomeningocele.

IMAGING

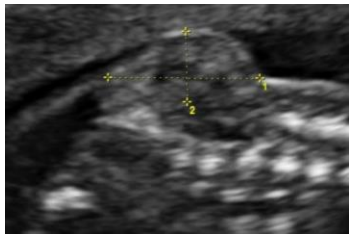

Figure 1 Sagittal image of fetal spine with overlying soft tissue mass measuring 19.2 x 8.9mm

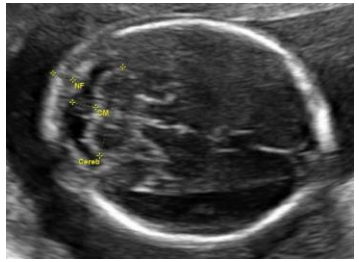

Figure 2 Axial brain – normal intracranial anatomy

CLINICAL PATHOLOGY

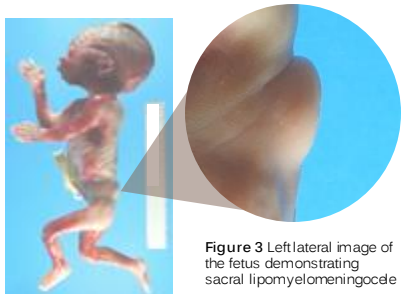

Figure 3 Left lateral image of the fetus demonstrating sacral lipomyelomeningocele

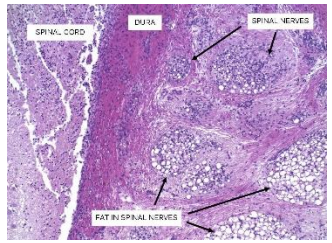

Figure 4 Histopathology showing fatty infiltration of spinal nerves

DISCUSSION

Closed spina bifida has been less widely studied than open types. Ultrasound findings are often more subtle than those seen in open spina bifida [1].

Lipomyelomeningocele is generally detected during the second-trimester morphology scan [2]. Characteristic findings include soft tissue lumbosacral mass with sacral extension of the spinal cord suggestive of tethering [3].

Most children born with lipomyelomeningocele will be symptomatic with neurogenic bowel or bladder at 6 months of life [4]. Symptoms of neuromuscular developmental delay may become apparent in early childhood.

Surgical intervention can delay development of symptoms, however retethering of the spinal cord may necessitate repeat surgeries [5].

CONCLUSION

This case outlines the antenatal diagnosis of closed spina bifida. Fetal MRI provides further diagnostic information in fetal life, which can guide counselling. Multidisciplinary team care is beneficial in supporting families. Early onset of spinal cord tethering is associated with earlier development of neurological symptoms.

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