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CASE REPORT

Infantile Presentation of Lipomyelocele: A Rare Case with MRI–Histopathology Correlation

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ABSTRACT

Background: A lipomatous mass that grows into the spinal canal and tethers the spinal cord is known as lipomyelocoele, a rare form of closed spinal disorder. Early detection is crucial to preventing progressive neurological impairment.

Case Summary: We present a case of a 4-month-old girl who, upon clinical examination, had a congenital lumbosacral swelling that was soft, mobile, and non-tender. A low-lying tethered cord that extends to the L5–S1 level was identified by an MRI. This cord was associated with spina bifida and the herniation of intact covered with skin neural components. Surgical excision was carried out on the patient. Histopathology confirmed the lipomyelocoele diagnosis.

Discussion: It helps to distinguish lipomyelocoele from dermoid/epidermoid cysts, myelomeningocele, and lipomyelomeningocele. Those having open neural tube defects, the lesion is covered in skin and does not have significant quantity of CSF. Rapid surgical intervention decreases neurological complications, and histopathology and MRI together are diagnostic.

Conclusion: This study highlightds how essential it is to mix pathological, radiological, and clinical findings because diagnosing lipomyelocoele. By decreasing complications related to tethered cords, early surgical intervention offers a favorable prognosis.

KEYWORDS: *Spinal developmental disorders, Lipomyelocoele, Spina bifida, Tethered cord, Paediatric neurosurgery.*

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INTRODUCTION

Lipomyelocele is a rare type of closed spinal dysraphism characterized by a lipomatous mass within the spinal canal, where the cord–lipoma interface lies at the dorsal defect. The lesion is skin-covered and does not involve meningeal herniation.[1] Population data suggest lipomatous spinal malformations, including lipomyelocele, occur in approximately 0.3–0.6 per 10,000 live births (≈3–6/100,000), making it much less common than open neural tube defects.[2,3] Precise figures for lipomyelocele alone are limited, but it is consistently described as rare.[4]

It arises from abnormal secondary neurulation and defective dysjunction, whereby incomplete separation of cutaneous and neural ectoderm permits mesenchymal infiltration and differentiation into adipose tissue.[1,5] Most cases are detected in infancy or early childhood, often due to lumbosacral swelling, cutaneous stigmata, or neurological and urological dysfunction.[4] Many infants are asymptomatic at birth but later develop tethered cord features.[2,3] A slight female predominance has been reported in some series.[2,4]

Clinically, lipomyelocele places children at risk of progressive neurological decline if untreated.[3] Early MRI-based detection and surgical detethering are crucial to prevent long-term morbidity.[1,2] Although retethering can occur in up to 10–20% of cases,[3] malignant transformation has not been reported, and the lesion is regarded as a benign congenital malformation.[1,3]

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CASE SUMMARY

Since birth, Baby Anabiya, a 4-month-old girl, has had swelling over her lumbosacral area. The swelling was soft, non-tender, and mobile on clinical examination. At presentation, there were no neurological problems.

Magnetic resonance imaging (MRI)

MRI of the lumbosacral spine revealed a low-lying tethered cord extending to the S1– level, with widening of the posterior elements of L4, L5, and S1 vertebrae, suggestive of spina bifida. A hyperintense fat signal with herniation of neural elements was noted, covered by intact skin. The sac measured 12 × 10 × 19 mm. The findings were consistent with spina bifida with lipomyelocoele and low-lying tethered cord. The patient underwent surgical excision of the swelling.

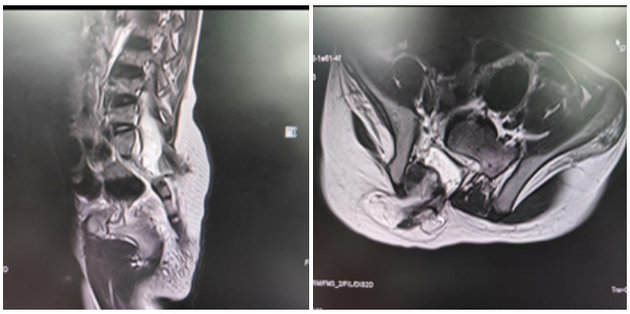

Figure 1: Presurgical MRI reveals a low-lying tethered cord extending to the S1– level, associated with spina bifida and herniation.

Figure 2: Gross Image showing gray white to gray brown solid areas.

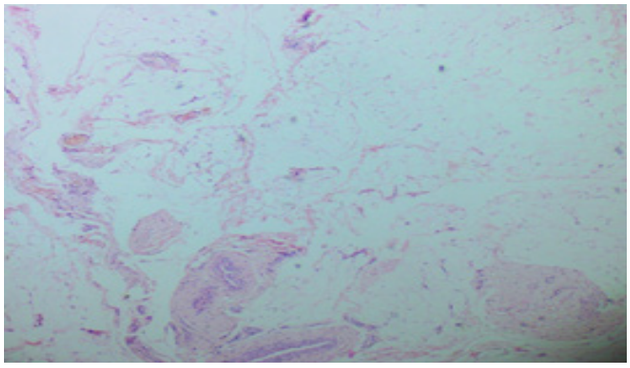

Figure 3: (H&E 10X)-showing lobules of mature adipocytes with fibrocollagenous tissue, congested blood vessels and few bundles of nerve fibers.

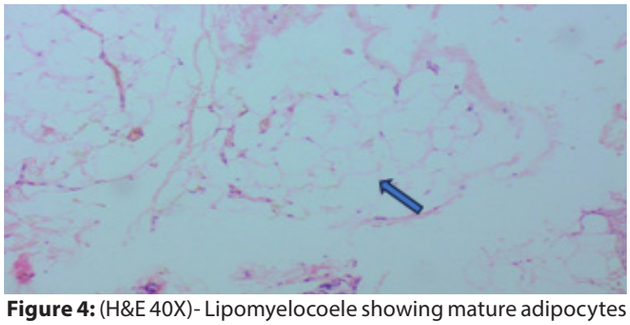

Figure 4: (H&E 40X)- Lipomyelocoele showing mature adipocytes with eccentrically placed nuclei and clear vacuolated cytoplasm.

Gross

Grossly the specimen consisted of multiple soft tissue fragments, grey-white to grey-brown in appearance, measuring 2 × 2 cm in aggregate. All tissue was processed.

Microscopy

Histological sections revealed lobules of mature adipocytes with eccentrically placed nuclei and clear vacuolated cytoplasm. Fibrocollagenous tissue, fibrofatty stroma, and congested blood vessels were present. Few bundles of nerve fibers were seen, showing a central axon surrounded by thin endoneurium. No granulomas, atypical, or malignant cells were identified. The histopathological impression was consistent with lipomyelocoele.

DISCUSSION

Lipomyelocoele is a form of closed spinal dysraphism resulting from defective neural tube closure and abnormal mesenchymal differentiation.[7] The fatty tissue extends into the spinal canal, often tethering the spinal cord and predisposing to progressive neurological impairment.[8]

Patients typically present with a lumbosacral swelling, which may or may not be associated with neurological deficits at birth.[9] Over time, tethered cord syndrome may manifest as motor weakness, sensory deficits, or bladder dysfunction.[10] MRI is the imaging modality of choice, as it demonstrates the fat signal intensity and the relation of neural tissue.[11]

Histopathology confirms the diagnosis by showing mature adipose tissue admixed with fibrous tissue and neural elements, as seen in our case. It must be distinguished from other lumbosacral lesions in infants. The main differentials include:

Lipomyelomeningocele – differs from lipomyelocoele by the presence of a CSF-filled sac protruding through the defect, in addition to fat and neural tissue.[10] In contrast, lipomyelocoele usually lacks a large CSF component and remains more localized under intact skin.

Myelomeningocele – an open neural tube defect with exposed neural placode and absent overlying skin, unlike lipomyelocoele which is covered by intact skin.[6,7]

Dermoid/Epidermoid cysts – show cystic components with keratin or dermal appendages, but lack the adipose–neural admixture seen in lipomyelocoele. [11]

The mainstay of treatment is early surgical excision with untethering of the cord to prevent progression of neurological sequelae. Early management improves the prognosis.[12]

CONCLUSION

This case highlights the importance of combining clinical, radiological, and histopathological evaluation in diagnosis of lipomyelocoele. Early surgical intervention is important to prevent tethered cord-related neurological deficits. Our patient was diagnosed early and underwent surgery, it highlights the value of timely multidisciplinary management in such congenital spinal dysraphisms.

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