Section: Pediatric Surgery

Case Report

LIPO-MYELOMENINGOCELE ASSOCIATED WITH CEREBRAL MIDLINE ANOMALIES, NEUROCUTANEOUS ALBINISM, AND UNILATERAL LOWER LIMB HYPOPLASIA: A RARE CASE REPORT

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ABSTRACT

Lipo-myelomeningocele is a rare form of closed spinal dysraphism that may occur in association with abnormalities of the central nervous system (CNS) and other organ systems. We report an exceptionally rare combination of findings in a three-month-old female infant presenting with lipo-myelomeningocele associated with agenesis of the corpus callosum, absence of the cavum septum pellucidum, neurocutaneous albinism, and hypoplasia of the left lower limb. This unusual constellation of anomalies suggests a complex developmental disturbance and highlights the importance of comprehensive neuroimaging evaluation and multidisciplinary management in infants with spinal dysraphism.

Keywords: Lipo-myelomeningocele, spinal dysraphism, cavum septum pellucidum, neurocutaneous albinism, neural tube defects.

INTRODUCTION

Lipo-myelomeningocele is a developmental anomaly in which adipose tissue remains continuous with the spinal cord through a posterior vertebral defect, resulting in tethering of neural elements.[1,2] Unlike open neural tube defects, the lesion is covered by intact skin and may not produce neurological symptoms early in life.[3] However, progressive neurological deterioration may occur as the child grows. Lipo-myelomeningocele commonly presents as an isolated malformation, although associated abnormalities involving the brain and musculoskeletal system have also been reported.[3,4,5] Agenesis of the corpus callosum is a disorder of cerebral commissural development and is frequently associated with other midline abnormalities.[6] Neurocutaneous albinism is characterized by hypopigmentation of the skin, hair, and eyes and may have associated neurological manifestations.[7] Limb hypoplasia has been described in complex congenital malformations and may reflect an early embryological developmental disruption.[5] Although LMMC has been reported in association with anomalies of the central nervous system and other organ systems, the coexistence of lipo-myelomeningocele, agenesis of the corpus callosum, neurocutaneous albinism, and limb hypoplasia is exceptionally rare. To the best of our knowledge, very few such cases with this combination of anomalies have been described in the literature.

Case Report

A three-month-old female infant was admitted from the outpatient department with a swelling over the lower back that had been present since birth. She was born at term by vaginal delivery to non-consanguineous parents following an uncomplicated pregnancy. There was no history of maternal illness, teratogenic drug exposure, or antenatal infection. Antenatal ultrasonography had been reported as normal. The neonatal period was uneventful except for difficulty in passing stools. On examination, a soft, non-tender lumbosacral swelling measuring 4 × 5 cm was noted. The swelling was covered with intact skin, with no evidence of ulceration or sinus opening. The right lower limb appeared normal, whereas the left lower limb was smaller in size, with decreased

muscle tone and grade II reflexes, suggestive of unilateral limb hypoplasia. In addition, the infant had generalized hypopigmentation of the skin and scalp hair, along with light-colored irises. Routine laboratory investigations were within normal limits. Magnetic resonance imaging (MRI) of the spine revealed a fat-containing lesion protruding through a posterior spinal defect and remaining continuous with the spinal cord, resulting in tethering of the cord. These findings were consistent with a diagnosis of lipo-myelomeningocele. [Figures 1 & 2]

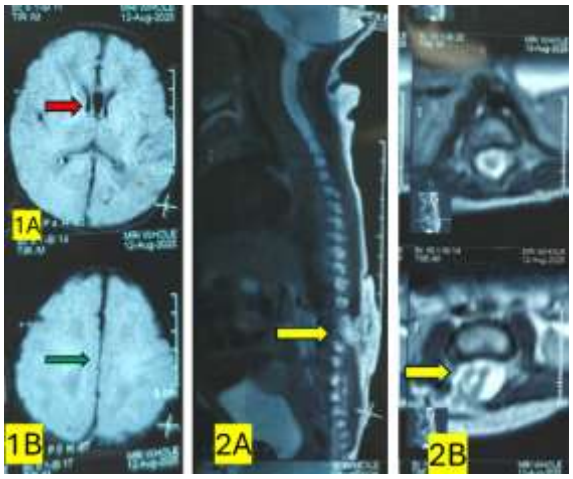

Figure 1A: MRI Scan of the brain showing Cavum septum pellucidum (red Arrow)
Figure 1B: MRI Scan of the brain showing Corpus Callosum agenesis (green arrow)
Figure 2A: MRI Scan of the spine sagittal section view showing Lipo-myelomeningocele (yellow arrow)
Figure 2B: MRI Scan of the spine, axial section view showing Lipo-myelomeningocele (yellow arrow)

Screening MRI of the brain revealed complete agenesis of the corpus callosum, associated ventricular configuration changes, and the presence of a cavum septum pellucidum. No evidence of hydrocephalus or other cortical malformations was noted. Ophthalmological examination showed reduced retinal pigmentation, supporting the diagnosis of neurocutaneous albinism. Based on the clinical and radiological findings, a final diagnosis of lipo-myelomeningocele associated with agenesis of the corpus callosum, cavum septum pellucidum, neurocutaneous albinism, and hypoplasia of the left lower limb was established. The patient was managed by a multidisciplinary team including pediatric surgeons, neurosurgeons, dermatologists, Dermatologists, Ophthalmologists and Plastic surgeons. A detailed pre-operative counselling regarding post-operative neurodevelopmental outcomes associated with agenesis of the corpus callosum and the need for long-term follow-up was provided to the parent. After pre-operative anaesthesia work-up, LMMC repair was done along with detethering of the spinal cord, and rotational flap skin cover was done. [Figure 2]

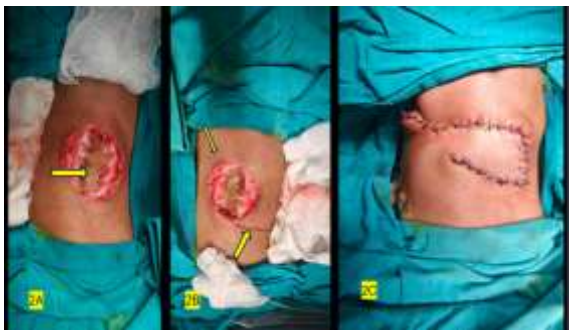

Figure 2: Intraoperative images during Lipo-myelomeningocele repair

Figure 2A: Intraoperative image showing the surgical field after excision of the lipomatous mass and dural closure, covered with a hemostatic mesh (yellow arrow).
Figure 2B: Intraoperative Image showing marking of rotational flap
Figure 2C: Intraoperative Image showing closure of flap

DISCUSSION

Lipo-myelomeningocele (LMMC) arises from a disorder of primary neurulation, specifically focal nondisjunction between the cutaneous ectoderm and neuroectoderm during early embryogenesis (3 to 4 weeks of gestational age). This allows mesenchymal adipose precursors to infiltrate the open neural placode, resulting in a subcutaneous lipomatous mass that maintains continuity with the spinal cord through a posterior dysraphism defect.[1,2] The lipoma typically tethers the cord, leading to progressive neurological compromise as the child grows, even if the infant is initially asymptomatic.[2] Unlike open neural tube defects (e.g., myelomeningocele), LMMC is covered by intact skin, which often delays diagnosis until cutaneous markers (e.g., swelling, hairy patch) or subtle deficits emerge.[3] Progressive tethered cord syndrome may manifest as bowel and bladder dysfunction, lower limb weakness, scoliosis, or orthopedic deformities, underscoring the need for vigilant monitoring and timely intervention.[2,8]
Isolated LMMC is the most common presentation, but associated anomalies involving the central nervous system (CNS), musculoskeletal system, and other structures have been described, albeit infrequently.[4,5,6] In LMMC intracranial abnormalities are rarely involved, while in open spinal dysraphism, Chiari II malformation, hydrocephalus, and corpus callosum anomalies are prevalent.[5] Brain anomalies such as agenesis of the corpus callosum (ACC) or absent cavum septum pellucidum (CSP) are exceptional in LMMC, as the embryological insult is typically localized to the spinal neurulation process rather than broadly affecting midline cerebral development.[5,6]
Agenesis of the corpus callosum results from failure of commissural plate formation and axonal migration across the midline (8 to 20 weeks of gestational age),

often co-occurring with other midline defects like absent CSP, which normally forms as the callosal fibers delineate the ventricular cavity.[6] The coexistence of LMMC with ACC and absent CSP in this case therefore suggests a more widespread early developmental disturbance, potentially involving disrupted signaling pathways (e.g., sonic hedgehog or Wnt) across multiple embryonic layers.[5] This broader midline vulnerability may explain the rarity of such combinations in closed dysraphism, where intracranial involvement is reported infrequently compared to open defects.[5,6]

The unilateral lower limb hypoplasia observed here further highlights the multisystem nature of the developmental insult. Limb anomalies in spinal dysraphism typically stem from chronic tethering, asymmetric neural involvement, or early mesodermal disruption, leading to leg length discrepancy, weakness, atrophy, or orthopedic deformities.[5] In LMMC, such features are often bilateral and progressive; unilateral presentation, as seen in this infant, is uncommon and supports an asymmetric embryonic disturbance affecting somites, neural crest derivatives, or vascular supply to the limb bud.[5] This finding reinforces the concept of a complex, multilayered embryopathy rather than isolated spinal pathology.

Neurocutaneous albinism, e.g., oculocutaneous albinism (OCA), adds another layer of complexity. OCA is a genetically heterogeneous disorder of melanin biosynthesis, characterized by hypopigmentation of skin, hair, and eyes, with associated ocular findings (e.g., foveal hypoplasia, nystagmus, reduced visual acuity).[7] While primarily dermatological and ophthalmological, OCA has recognized neurological associations, including abnormal visual pathway decussation and potential subtle CNS involvement.[7] The concurrence of OCA with LMMC and cerebral midline anomalies is extraordinarily rare, with no prior reports of this specific triad. This suggests a possible shared genetic or epigenetic mechanism disrupting neural crest derivatives (which contribute to pigmentation) and neurulation processes. The presence of generalized hypopigmentation and light irises in this case, confirmed by ophthalmological evaluation, emphasizes the need for thorough systemic assessment in atypical spinal dysraphism presentations.

The constellation of LMM, ACC with absent CSP, unilateral limb hypoplasia, and neurocutaneous albinism represents an unprecedented combination, to the best of our knowledge very few reported instances in the literature. This rarity underscores the importance of comprehensive neuroimaging (whole-spine and brain MRI) in infants with spinal dysraphism, even when asymptomatic, to detect occult CNS anomalies that may influence neurodevelopmental prognosis.[4,5,6] ACC is associated with variable outcomes, ranging from normal cognition to intellectual disability, epilepsy, or behavioral challenges, often modulated by coexisting hydrocephalus or cortical malformations (absent here).[6] Early identification facilitates proactive monitoring and intervention.

Management of LMMC focuses on preventing progressive tethered cord deficits through surgical detethering and lipoma resection. Current evidence favors early intervention (ideally within the first year of life) to minimize irreversible neurological damage, as delayed surgery increases risks of bowel/bladder dysfunction, motor deficits, and orthopedic complications.[8,9] Techniques such as complete lipoma removal, watertight dural closure, rotational flap coverage, and post-operative prone position have been shown to reduce risks.[8,9] Post-surgical outcomes vary, with complications (e.g., CSF leak, infection, wound issues, new deficits) reported in up to 42.5% of cases, often linked to age at surgery, lipoma type, excision extent, and closure technique.[8] In this infant, early repair with detethering and flap cover aligned with these recommendations, aiming to halt progression while preserving function. Multidisciplinary involvement—pediatric neurosurgery, surgery, dermatology, ophthalmology, plastic surgery, and psychology—is essential for addressing the multisystem anomalies, pre-operative counseling on neurodevelopmental risks (particularly from ACC), and long-term follow-up for tethered cord surveillance, visual/developmental assessments, and rehabilitation.

In conclusion, this case illustrates a rare, multifaceted developmental disorder requiring heightened awareness of extraspinal associations in closed spinal dysraphism. Detailed neuroimaging, genetic evaluation (if feasible), and prompt multidisciplinary intervention offer the best chance for optimizing outcomes in such complex presentations.

CONCLUSION

This case represents an exceptionally rare association of lipo-myelomeningocele with cerebral midline anomalies, neurocutaneous albinism, and unilateral lower limb hypoplasia, necessitating comprehensive imaging and a multidisciplinary evaluation to identify associated anomalies and guide timely management for improved postoperative outcomes.

Conflict of interest: All the authors of this manuscript do hereby declare that we have no conflict of interest whatsoever in the case report mentioned in this manuscript.

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