Case study

Primary Neural Tube Defects in Pediatrics – A Focus on Lipomeningocele

ABSTRACT

Background: Lipomeningocele is a congenital abnormality of the neural tube. It affects approximately one in every 50,000 infants. This is one of the most uncommon varieties of Spina bifida, which happens when a neural tube does not shut completely and sticks out of the Spinal column, forming a sack beneath the skin. During embryonic development, about day 21 or week 3, neural folds fuse to form a neural tube and form a complete neural tube on the 28th day. The unfused part of the spinal cord leads to Spina bifida. Getting enough folic acid, during pregnancy can help to prevent neural tube defects. Mothers who are obese, have poorly controlled diabetes, and mothers who take certain anti-seizure medicines are at more risk of having a baby with a neural tube defect. **Case Presentation:**A 6-month-old male child presented with the chief complaints of swelling in the sacral region since birth, non-progressive in size, and was not reducible. Magnetic resonance imaging of the lumbar spine revealed that there is Lipomeningocele at the sacral region and low insertion of the spinal cord with tethering. **Discussion:**The primary objective is to reduce the stretching or straining of the spinal cord that occurs as the child grows. Prompt surgical interventions are crucial for the prevention of neurological deterioration. **Conclusion:**A neurosurgeon works to "untether" the spinal cord, separating the spinal cord from the back tissues. The idea is to reduce the stretching or straining of the spinal cord that occurs as the child grows.

Keywords:*Lipomeningocele, Spina bifida, Neural tubedefect, neurological deterioration, untethering, cerebrospinal fluid,Magnetic Resonance Imaging.*

INTRODUCTION

Lipomeningocele is a congenital lesion associated with spina bifida. Also known as a type of closed spinal dysraphism.Spinal dysraphism encompasses congenital problems that result in an abnormal bony formation of the spine and the spinal cord. This congenital pathology is caused by the maldevelopment of the ectodermal, mesodermal, and neuroectodermal tissues[1]. The spina bifida is a congenital abnormality that results from incomplete development of the neural tube. It is commonly used as a nonspecific term referring to any degree of neural tube closure. Spinal dysraphisms can be classified as either open or closed dysraphisms.

Open spinaldysraphismsincludemeningocele,myelomeningocele,myeloschisis,encephalocele, andanencephaly. Allinvolveexposureof nervous tissue and/or meninges to theexternalenvironment. Closed spinaldysraphismssuch as Lipomeningocele, lipomyelomeningocele,diastematomyelia,and Spina Bifida occultahavenoexposedneuraltissue and areaccompaniedbycutaneous markers and include lesions such as subcutaneous masses, capillary hemangioma, dimples, and hairy nevus. Itmight develop through a Spina bifida oralong the midline of the calvaria[2].It consists of fat-filled meningeal tissue covered by skin that is continuous with the flax cerebri in the head and the dura of the spinal cord in the vertebral column. Lipomeningocelerepresents a rarebut complexneurologicaldisorder thatmay present with neurologicaldeteriorationsecondarytoan inherent tetheredspinal cord.

Neural tube defects (NTDs) are among the leading noninfectious birth defects with a worldwide prevalence of 1–2 per 1000 live births causing significant infant morbidity and mortality and known to have multifactorial-polygenic origin for occurrence where both genetic and environmental factors including maternal nutrition are reported to have considerable contributions. The prevalence of NTDs in India has been reported are range from 0.5 to 11 per 1,000births. The prevalence of Lipomeningocelehasbeenfoundto range between 0.3 and0.6per 10,000 livebirths. Spinal dysraphisms are caused by the disruptions in the embryogenetic cascade occurring during early embryonic development between gestational weeks 2 and 6.

Severaltypes of closedspinal dysraphismsresultfromembryological abnormalitiesduringprimaryneurulation[3]. Thosethatarisefrompremature disjunctionresultin the fusionofthespinalcordwithfattyelements, themost common of which is a lipomeningocele. When premature disjunction occurs, the epithelial ectoderm separates prematurely from the neural ectoderm, allowing mesenchyme to contact the inner portion of the developing neural tube. The dorsal surface of the closing neural tube induces mesenchyme to form fat, and this prevents proper neurulation.

Theextentofthefattytissueislimited laterally by the neural ridge because the ventral surface of the neural plate induces the mesenchyme to form meninges[4]. This results in a junction between meninges and fat at the neural ridge, andthus the lipoma extends posteriorly through the meningeal and bony defect and into subcutaneous tissues in the extradural space. The neural placode-lipoma interface, which is the connection between the spinal cord and the lipoma, can lie outside of, within, or at the edge of the spinal canal. Lipomeningocele is characterized bya placode-lipoma interface located outside the spinal canal. A tethered cord is inherently associated with Lipomeningocele as the lipoma tethers the cord to the adjacent dura and soft tissue.

This condition of Lipomeningocele develops due to inadequate levels of folate (vitamin B9) and vitamin B12 during pregnancy[5]. The likelihood of having another child with the same issue is somewhat higher in couples who have already had one child with a neural tube defect.

Some anti-seizure medications, such as Valproic acidand Immunosuppressant like Methotrexate may cause neural tube abnormalities when taken during pregnancy. This might happen because they interfere with the body's ability to use folate and folic acid. Women with diabetes who don't have well-controlled blood sugar have a higher risk of having a baby with spina bifida.Pre-pregnancy obesity is associated with an increased risk of neural tube birth defects. Lipomeningoceleis characterized by a subcutaneous lipoma that is generally located in the lumbar or sacral region[6].

Thesubcutaneouslipomaextendsthrough a defectinthe lumbo dorsal fascia, vertebral neural arch, and dura, attaching to an elongated and tethered spinal cord.The most common presenting symptom is a fatty mass positionedin the midline or just off the midline in the lumbosacral region.Additionally, the majority presentwithother skin lesionsassociatedwith the lipoma, including a hairynevus, skindimples, andcutaneoushemangiomas. Loss of neurological function has been found to increase with age because of progressive conus tethering and injury to nervous tissue.Loss of motor and sensory functions.Thedisease progression can result in frequent urinary tract infections and neurogenic bladder and bowel incontinence or constipation, as well as leglength discrepancy, foot deformities, gait abnormalities, scoliosis, spasticity, and back and leg pain, complete paralysis of bowel and bladder[7].

It can be diagnosed prenatally by a 3D ultrasonography, by examination a well-demarcated subcutaneous mass can be detected in the lower sacral area at 36 weeks. The spinal cord was observed to extend into the sacral area instead of being located in the upper lumbar spine, and an additional echogenic intra-spinal mass contiguous with the lower spinal cord can be identified.MRI hasaidedinboththediagnosisandtreatment of Lipomeningocele. The treatment includes surgical intervention by which a neurosurgeon, on the other hand, works to "untether" the spinal cord that is separating the spinal cord from the back tissues[8]. The idea is to reduce the stretching or straining of the spinal cord that occurs as the child grows. Prompt surgical interventions are crucial for the prevention of neurological deterioration.

PRESENTATION OF THE CASE

A 6-month-oldmale child presented to the pediatric wardwith the chief complaint of swelling in the sacral region since birth that wasnot associated with pain, non-progressive in size, and was not reducible.During the general examination, the blood pressure is 110/60mmHg, the Pulse rate is 128/min, and the temperature is 97°F. Laboratory investigations observed were as follows; Serum Sodium:142 mEq/L, Serum potassium: 5.2 mEq/L, Serum Creatinine: 0.7 mg/dL), BloodUrea: 16 mg/dL, Serum Calcium: 9.4 mg/dL), Hemoglobin: 10.2 g/dL,PCV:28.7%,RBC: 3.1 Millions/Cu.mm,WBC: 13,100 cells/Cu.mm), Neutrophils:80%, Lymphocytes:14%, Eosinophils: 1%, Monocytes 5%, MCV: 93.4 fL.MRI of the Lumbar Spine shows that there is a CSF intensity lesion at the posterior aspect of $S_4$and S_5 and is communicating with the Thecal Sac suggestive of a Meningocele (approx. 2.0*3.0*0.7 cm). There is a cap like T_1, and T_2hyperintensity suggestive of fat component, at the posterior aspect of meningocele. The thickness of the fat component is about 2.2 cm and 2.2 cmcraniocaudal. There is low insertion of the spine cord with tethering noted at the posterior Thecal sac at the region of the neck of the meningocele.

Impression: Lipomeningocele at the sacral region, and low insertion of the spinal cord with tethering, suggested clinical correlation. The treatment that has to be given before the surgical intervention to prevent various infections includesInj. Ceftriaxone 200mg IV STAT, and Inj. Amikacin 35mg IV STAT. The medicines given after the Surgery were Syrup. Taxim-O 1ml oral Two Times a day, Syrup. PCM 1-2ml oral/Three Times a Day, Inj. Ceftriaxone 4ml IV Two Times a Day and Inj.Amikacin 50mg IV/at night time.

DISCUSSION

Lipomeningocele is typically diagnosed at birth and post-natal manifests as a subcutaneous lipoma over the lower back and is associated with tethered cord syndrome (TCS).In our case, the patient had been presented with swelling in the sacral region since birth and it was not associated with pain, Non-progressive in natureand was not reducible. MRI revealed that there is a Lipomeningocele at the sacral regionand lower insertion of the spinal cord with tethering[9].

Tethering of the spinal cord has been associated with a various range of pathological conditions which includes a thickened and constricting filum terminal, intradural lipomas with or without accompanying extradural components, intradural fibrous adhesions, and the presence of myelomeningocele.This condition should be surgically treated before it progresses into neurological symptoms, such as weakness, tingling sensation, and pain in the lower limbs[10].

The time of surgical intervention is crucial, it is recommended that the surgery should be performed early or prior to the presence of neurological symptoms or neurological deterioration. In order to treat Lipomeningocele with tethered cord syndrome (TCS) surgically, the lipomeningocele must be repaired, the spinal cord must be decompressed and the tether must be carefully detached. In contrast, intra-durallipomas like the one in this instance bind the cord where they enter the dura[11]. To separate the intra-dural and extra-dural components, the dura must beopened. Intra-dural lipomatous mass should be decompressed and the cord should be untethered. The part of the mass adhered to the Conus was left in situ to prevent injury to the neural tissue[12]. A water-tight closure of the dura was achieved.

It can be difficult to control the tethering effects of shorter nerve roots or arachnoid adhesions. There may be additional treatments or long-term management needed if complications such as recurring tethering, CSF leaks, infections, neurological impairments, and scar tissue formation occur.The primary objective of the surgery is consistent which is to prevent future or further neurological deterioration and preserve or improve current neurological function[13].

CONCLUSION:

Lipomeningocele is a form of closed neural tube defect with unknown predisposing factors. Due totheriskofworseningneurologicalandurologicalfunction secondary to a tethered spinal cord, it is nevertheless crucial to recognize this condition for timely intervention, so that prompt treatment can be given since a tethered spinal cord increases the risk of poor neurological and urological functions.Magneticresonanceimagingand neurophysiological testing are useful tools for identifying spinal cord pathology and assisting with surgical planning. Because bowel and bladder paresis remains the primary morbidity of this disease, early intervention either prior to symptom developmentor at the first onset ofsymptoms is recommended to improve post-operativeoutcomes.The surgery and antibiotics are the main therapeutic options used to reduce infections that could be avoidedand the surgery must be performed before the symptoms get worseso that the patient gets recovered and discharged.

CONSENT

All authors declare that written informed consent was obtained from the patient's Parent, as the patient is a 6-month-old Infant and the signed Consent Form is submitted along with the manuscript.

References:

1.Donovan MF, Bordoni B. StatPearls [Internet]. StatPearls Publishing; Treasure Island (FL): Feb 2, 2022. Embryology, Yolk Sac. [PubMed]

2.Neural Tube DefectsAuthor manuscript; available in PMC 2015 Jul 1.*Published in final edited form as*Annu RevNeurosci. 2014; 37: 221–242.doi: 10.1146/annurev-neuro-062012-170354PMCID: PMC4486472EMSID: EMS63689PMID: 25032496.

3. Meuli M, et al. The spinal cord lesion in human fetuses with myelomeningocele: Implications for fetal surgery. *J Pediatr Surg.* 1997;32: 448–452. [PubMed] [Google Scholar].

4. Sharifah A. Othman, Abdulrazaq AlOjan, Mohammed AlShammari, and Ahmed Ammar. Awareness of spina bifida among the family of an affected child. Saudi Med J. 2019; 40(7): 727-731. PMCID: PMC6757207. PubMed, PMID: 31287135.

5. Tamara Richards, Jamal Brown. Folate Deficiency and Neural Tube Defects in Pregnancy. US Pharm. 2021;46(9):HS-8-HS-12.

6. Joslyn Gober, Sruthi P. Thomas, and David R. Gater. Pediatric Spina Bifida and Spinal Cord Injury. J Pers Med. 2022 Jun; 12(6): 985. PMCID: PMC9225638. PubMed, PMID: 35743769.

7. Total Resection of Complex Spinal Cord Lipomas. Neurol Med Chir. 2015 Sep; 55(9):695-721. PMCID: PMC4605078. PubMed, PMID: 26345666.\

8. Feldman RP, Marcovici A, Lasala PA: Intracranial lipoma of the sylvian fissure: case report and review of the literature. *J Neurosurg* 94: 515-519, 2001

9. Dias MS, Partington M. Embryology of myelomeningocele and anencephaly. Neurosurg Focus. 2004;16(2):E1. doi:10.3171/foc.2004.16.2.2.

10. Adzick NS, Thom EA, Spong CY, Brock 3rd JW, Burrows PK, Johnson MP, et al. A randomized trial of prenatal versus postnatal repair of myelomeningocele. N Engl J Med. 2011;364(11):993–1004. doi:10.1056/NEJMoa1014379.

11. Tulipan N, Wellons 3rd JC, Thom EA, Gupta N, Sutton LN, Burrows PK, et al. Prenatal surgery for myelomeningocele and the need for cerebrospinal fluid shunt placement. J NeurosurgPediatr. 2015;16(6):613–20. doi:10.3171/2015.7.PEDS15336.

12. Clayton DB, Tanaka ST, Trusler L, Thomas JC, Pope JC, Adams MC, et al. Long-term urological impact of fetal myelomeningocele closure. J Urol. 2011;186(4 Suppl):1581–5. doi:10.1016/j.juro.2011.04.005.

13. Satar N, Bauer SB, Scott RM, Shefner J, Kelly M, Darbey M. Late effects of early surgery on lipoma and lipomeningocele in children less than 1 year old. J Urol. 1997;157(4):1434–7.

14. Kessler TM, Lackner J, Kiss G, Rehder P, Madersbacher H. Predictive value of initial urodynamic pattern on urinary continence in patients with myelomeningocele. NeurourolUrodyn. 2006;25(4):361–7. doi:10.1002/nau.20230.

15. Almodhen F, Capolicchio JP, Jednak R, El Sherbiny M. Postpubertal urodynamic and upper urinary tract changes in children with conservatively treated myelomeningocele. J Urol. 2007;178(4 Pt 1):1479–82. doi:10.1016/j.juro.2007.05.171.

16. Hopps CV, Kropp KA. Preservation of renal function in children with myelomeningocele managed with basic newborn evaluation and close follow-up. J Urol. 2003;169(1):305–8. doi:10.1097/01.ju.0000040590.35948.bc.

17. Dorward NL, Scatliff JH, Hayward RD. Congenital lumbosacral lipomas: pitfalls in analysing the results of prophylactic surgery. Childs Nerv Syst. 2002;18(6–7):326–32. doi:10.1007/s00381-002-0624-x.

18. Sarris CE, Tomei KL, Carmel PW, Gandhi CD. Lipomyelomeningocele: pathology, treatment, and outcomes. Neurosurg Focus. 2012;33(4):E3. doi:10.3171/2012.7.FOCUS12224.

19. Sebold CD, Melvin EC, Siegel D, Mehltretter L, Enterline DS, Nye JS, et al. Recurrence risks for neural tube defects in siblings of patients with lipomyelomeningocele. Genet Med. 2005;7(1):64–7. doi: 10.109701.GIM.0000151158.09278.2B.

20. Agopian AJ, Canfield MA, Olney RS, Lupo PJ, Ramadhani T, Mitchell LE, et al. Spina bifida subtypes and sub-phenotypes by maternal race/ethnicity in the National Birth Defects Prevention Study. Am J Med Genet A. 2012;158A(1):109–15. doi:10.1002/ajmg.a.34383.