สกิน ดิมเพิล เป็นมากกว่ารอยบุ๋ม

พงศ์ภัทร์ วรสายัณห์
โรงพยาบาลสมเด็จพระบรมราชเทวี ณ ศรีราชา สภากาชาดไทย

บทคัดย่อ

ไลโปมัยอีโลซีล (lipomyelocele) พบได้น้อยมากในทารกแรกเกิด พบได้ตั้งแต่ 0.3 – 0.6 ต่อทารกแรกเกิด 10,000 คน แพทย์ผู้ให้การรักษารายงานผู้ป่วยเด็กเพศหญิงอายุ 6 ปี ได้รับการวินิจฉัยเป็นไลโปมัยอีโลซีล โดยไม่พบอาการแสดงใดๆ ตั้งแต่แรกเกิดมาเป็นเวลา 6 ปี หลังจากนั้นเริ่มมีอาการผิดปกติทางระบบประสาท จากการทบทวนประวัติผู้ป่วยรายนี้ พบว่า มีรอยบุ๋มที่ผิวหนังบริเวณกลางหลังตั้งแต่แรกเกิดและไม่ได้รับการตรวจรักษา ตอนอายุ 7 ปี ผู้ป่วยได้รับการรักษาด้วยการผ่าตัดเอาก้อนไขมันออกและซ่อมแซมเยื่อบุดูรา แต่อย่างไรก็ตามการรักษานี้ไม่สามารถแก้ไขอาการผิดปกติทางระบบประสาทให้กลับคืนได้

คำสำคัญ: สไปนอล ดิสราฟิซึม ไลโปมัยอีโลซีล สกิน ดิมเพิล

ผู้นิพนธ์หลัก:
พงศ์ภัทร์ วรสายัณห์
ฝ่ายอายุรกรรม โรงพยาบาลสมเด็จพระบรมราชเทวี ณ ศรีราชา สภากาชาดไทย
อำเภอศรีราชา จังหวัดชลบุรี
อีเมล์: pongpatv@yahoo.com

Skin dimple, more than a pit

Pongpat Vorasayan

Division of Medicine, Queen Savang Vadhana Memorial Hospital, Thai Red Cross

Abstract

Lipomyelocele is an uncommon condition in neonates with a low prevalence of 0.3 – 0.6 per 10,000 live births. We report a case of 6-year-old girl with lipomyelocele who had no neurological deficit for 6 years. Afterward, she presented with progressive neurological deficits. Her history was revealed that she had a typical cutaneous marker which was abandoned since birth. Finally, she received a surgical removal of a lipomatous mass and a dural reparation at the age of 7 years old. Unfortunately, her neurological deficits cannot be recovered.

Keywords: spinal dysraphism, lipomyelocele, skin dimple

Corresponding author:

Pongpat Vorasayan

Division of Medicine, Queen Savang Vadhana Memorial Hospital, Thai Red Cross

Sriracha, Chonburi

E-mail: pongpatv@yahoo.com

Introduction

Neural tube defects are congenital anomalies of neurulation in embryogenesis with a wide range of clinical manifestations. Lipomyelocele is one of the neural tube defects which are classified in closed spinal dysraphism (occult spinal dysraphism). It usually presents with cutaneous markers or neurological deficits secondary to tethered spinal cord. Mainstay of treatment is surgery, which aim to prevent further neurological deficits. We report a case of delayed diagnosed lipomyelocele with review literatures in aspect of treatment and outcome.

Case report

A 6-year-old girl presented with a progressive foot drop on her right leg, numbness of her right foot and episodic urinary urgency for 6 months. Physical examinations revealed muscle strength of 3/5 of right tibialis anterior muscle, extensor hallucis longus muscle, and gastrocnemius muscle. She also had a hyporeflexia of the right ankle and decreased pin-prick sensation of the lateral aspect of the right foot. She had a skin dimple which located in the midline of lumbrosacral region, her mother claimed that it presented at birth.

Magnetic Resonance Imaging (MRI) of the lumbrosacral spine showed spina bifida occulta from L4 through S5 level. The corresponding spinal canal was capacious and posterior scalloping of L4 and L5 vertebrae were detected. A 2.2×1.6×6.2 cm subcutaneous fatty mass was seen at the covering skin that represents lipoma. It extended toward from the subcutaneous plane through the defects into the spinal canal. It inserted into the dorsal surface of the tethered spinal cord which terminated at upper S1 level (Figure 1).

She was diagnosed with lipomyelocele with no associated abnormalities. Removal of the lipomatous mass, untethering the spinal cord, and duroplasty with autologous fascial graft were done. After the operation, she had more weakness in the former described muscles, muscle strength of the affected limb become 1/5. She developed neurogenic bladder resulting from retained catheter since the operation.

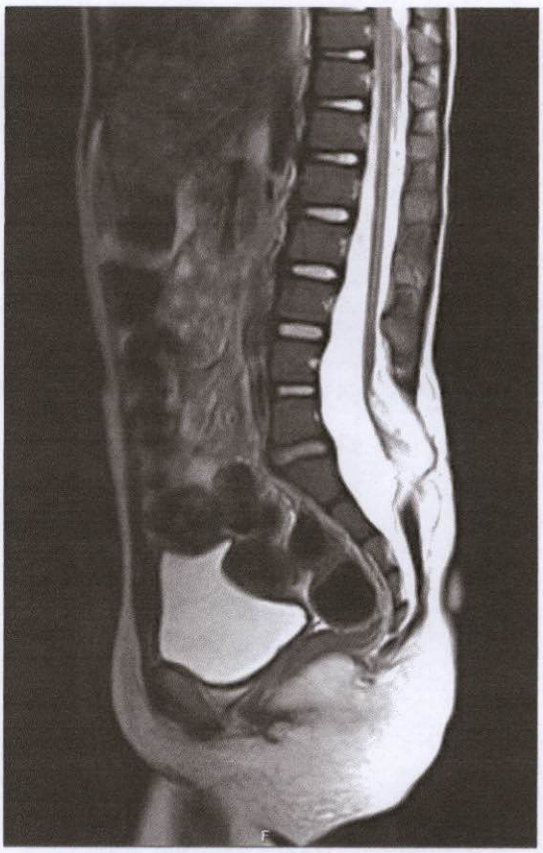

Figure 1 T2-weighted image showed lipomyelocele with tethered cord ending at upper S1 level.

Discussion

Neural tube defects can affect cranium, spine or both as in the following: 1. Cranial defects including anencephaly, exencephaly, and encephalocele, 2. Open spinal dysraphism (spina bifida aperta) is characterized by herniation of the nervous tissue through the defect in the spinal column including myelomeningocele and meningocele, 3. Closed spinal dysraphism (spina bifida occulta) is characterized by unexposed nervous tissue. Lipomyelocele is a common subtype of the closed spinal dysraphism. The prevalence of all types of spinal dysraphism occur at a frequency of 0.5 to 8 per 1,000 live births[1] while the prevalence of lipomyelocele has been range from 0.3 to 0.6 per 10,000 live births[2-4].

Risk factors of spinal dysraphism are ethnic groups, nutritional deficiency, and maternal diabetes. First, prevalence of spinal dysraphism in Hispanic group is 3.26 per 10,000 live births while prevalence of spinal dysraphism in non-Hispanic whites and non-Hispanic blacks are 2.57 and 2.07 per 10,000 live births, respectively[2]. Second, folic acid deficiency had been reported as a strong association with the open spinal dysraphism[5]. There are also several reports of a significant higher prevalence of open spinal dysraphism in neonates which born to pregnant women who take certain antiepileptic drugs which effect in folic acid metabolism[6-8]. Third, maternal diabetes has been associated with increased incidence of the syndrome of caudal regression without exactly known mechanism. Conversely, there are no significant risk factors which closely correlated with spinal dysraphism.

Lipomyelocele results from abnormal embryogenesis during primary neurulation. According to premature dysjunction of neuronal ectoderm and epidermal ectoderm, these abnormal development allow mesenchyme to attach to the developing neural tube[9]. This mesenchyme is induced by dorsal surface of neural tube to form fatty tissue, and prevents proper neurulation.

Closed spinal dysraphism are commonly accompanied by cutaneous markers such as subcutaneous masses, tails, capillary hemangioma, dimples, and hairy nevus in 43-95% of cases[10,11]. In Natarajan cases series reported that merely 40% of patients with lipomas without a dural defect had cutaneous markers while all of patients with lipomas with dural defect had cutaneous markers[12]. These cutaneous markers can be used as indicators for further investigation in asymptomatic neonates.

Progressive neurological deficits, urinary deficits, orthopedic deformities and the sequelae of tethered cord has been found to increase with age because of progressive clonus tethering and injury to nervous tissue[13-15]. Hoffman et al. observed that 62.5% of patients were neurologically asymptomatic before 6 months of age while only 29.3% of patients who presented after 6 months of age were asymptomatic[14]. Among the symptomatic patients, they can present as weakness or hypalgesia of one or both lower extremities, gait abnormalities, sphincter disturbance, scoliosis, limb length discrepancy, foot deformities, or back and leg pain.

Treatment strategies of lipomyelocele mainly are surgical interventions and rehabilitation in deficit cases. Surgery in these groups of patients consisted of removal of lipomatous mass, avoid injuring any nerve roots or clonus medullaris that might course close to dural defect, and primary dural closure or duroplasty in large defects[12,16]. Complications can be divided into two types: wound complications and neurological complications. Incidence of wound complications occurred in 10-30% of patients included wound infection, flap necrosis, and CSF leakage[17,18]. Neurological deterioration after surgery can be transient or permanent neurological deficits. Temporary weakness or urinary deficits reported in 3.4-7.5% after the operation[12,19]. Permanent neurological deficits has been found 3-10% which could be influenced by surgical treatment, incomplete release of tethering, or retethering[12,20]. Retethering occurred 3.45-20% of the patients after follow-up for 3-8 years[17]. Treatment outcomes in one case reported that 44.83% of patients improved, 18.97% stabilized deficits, 3.45% deteriorated, and 32.76% of patients were asymptomatic, same as presentation[12]. Preservation or improvement of neurological outcomes is better when patients presented and underwent surgery before 2 years of age[12].

In conclusion, we report a case of lipomyelocele which was diagnosed by presentation of neurological deficits. Despite this patient had a typical cutaneous marker at birth, which we; as a physician, should aware and promptly investigate for spinal dysraphism. We had missed a 6-year-peroid of a curative treatment for her. Our purpose is to emphasize the important of a skin dimple in neonate is not just a pit.

References

1. Saitsu H, Yamada S, Uwabe C, et al. Development of the posterior neural tube in human embryos. Anat Embryol 2004;209:107-17.
2. Agopian AJ, Canfield MA, Olney RS, 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:109-15.
3. De Wals P, Van Allen MI, et al. Impact of folic acid food fortification on the birth prevalence of lipomyelomeningocele in Canada. Birth Defects Res A Clin Mol Teratol 2008;82:106-9.
4. Forrester MB, Merz RD. Descriptive epidemiology of lipomyelomeningocele, Hawaii, 1986-2001. Birth Defects Res A Clin Mol Teratol 2004;70:953-6.
5. Betteridge KJ. Comparative aspects of conceptus growth: a historical perspective. Reproduction 2001;122:11-9.
6. Ceylan S, Duru S. Valproic acid sodium-induced spina bifida occulta in the rat. Neurosurg Rev 2001;24:31-4.
7. Morrow J, Russell A, Guthrie E, et al. Malformation risks of antiepileptic drugs in pregnancy: a prospective study from the UK Epilepsy and Pregnancy Register. J Neurol Neurosurg Psychiatry 2006;77:193-8.
8. Rosa FW. Spina bifida in infants of women treated with carbamazepine during pregnancy. N Engl J Med 1991;324:674-7.
9. Warder DE. Tethered cord syndrome and occult spinal dysraphism. Neurosurg Focus 2001;10:e1.
10. Gibson PJ, Britton J, Hall DM, et al. Lumbosacral skin markers and identification of occult spinal dysraphism in neonates. Acta Paediatr 1995;84:208-9.
11. Guggisberg D, Hadj-Rabia S, Viney C, et al. Skin markers of occult spinal dysraphism in children: a review of 54 cases. Arch Dermatol 2004;140:1109-15.
12. Muthukumar N. Congenital spinal lipomatous malformations: part II--Clinical presentation, operative findings, and outcome. Acta Neurochir 2009;151:189-97.
13. Hoffman HJ, Hendrick EB, Humphreys RP. The tethered spinal cord: its protean manifestations, diagnosis and surgical correction. Childs Brain 1976;2:145-55.
14. Hoffman HJ, Taecholarn C, Hendrick EB, et al. Management of lipomyelomeningoceles. Experience at the Hospital for Sick Children, Toronto. J Neurosurg 1985;62:1-8.
15. Kanev PM, Bierbrauer KS. Reflections on the natural history of lipomyelomeningocele. Pediatr Neurosurg 1995;22:137-40.
16. Sarris CE, Tomei KL, Carmel PW, et al. Lipomyelomeningocele: pathology, treatment, and outcomes. Neurosurg Focus 2012;33:E3.
17. Arai H, Sato K, Okuda O, et al. Surgical experience of 120 patients with lumbosacral lipomas. Acta Neurochir 2001;143:857-64.
18. Kanev PM, Lemire RJ, Loeser JD, et al. Management and long-term follow-up review of children with lipomyelomeningocele, 1952-1987. J Neurosurg 1990;73:48-52.
19. Pierre-Kahn A, Zerah M, Renier D, et al. Congenital lumbosacral lipomas. Childs Nerv Syst 1997;13:298-334.
20. James HE, Williams J, Brock W, et al. Radical removal of lipomas of the conus and cauda equina with laser microneurosurgery. Neurosurgery 1984;15:340-3.