Case Report

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20160176

Case of congenital diastematomyelia type 1: a rare presentation

Balaji M. D., Shivaprakash N. C., Prashanth Kumar Thangella*

Department of Pediatrics, Adichunchanagiri Institute of Medical Sciences, B.G nagara, Nagamangala taluk, Mandya District, Karnataka, Pincode -571448, India

Received: 19 November 2015 Revised: 21 December 2015 Accepted: 09 January 2016

*Correspondence:

Dr. Prashanth Kumar Thangella, E-mail: prashanth.tpr@gmail.com

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ABSTRACT

Diastematomyelia is a rare congenital anomaly that results in the splitting of the spinal cord in a longitudinal (sagittal direction). It is also known as a split cord malformation, refers to a type of spinal dysraphism (spina bifida occulta) when there is a longitudinal split in the spinal cord. We report a case of new born female baby with Diastematomyelia type 1 associated with low lying conus, tethered cord and dorsal dermal sinus.

Keywords: Diastematomyelia, Tethered cord, MRI Scan

INTRODUCTION

Diastematomyelia, also known as Split Cord Malformation (SCM) is a congenital spinal anomaly in which there is longitudinal splitting of the spinal cord. The two hemicords may be separated by a fibrous, bony or cartilaginous septum.1 Antenatal detection of cord abnormalities by USG can be limited by acoustic shadowing from the spine, fetal position and the amount of liquor, though it is usually quite accurate in the diagnosis of spina bifida, in experienced hands, when correlated with maternal alpha-fetoprotein levels.² The mean (range) gestational age for diagnosis of Diastematomyelia was 21 (13-25) weeks. The main sonographic findings were widening of the spinal canal in the coronal view and an additional echogenic focus in the posterior part of the spinal column in the axial view and associated elevated levels of amniotic fluid α-fetoprotein (AF-AFP) and acetylcholinesterase (AF-AChE) in many

MRI is a safe and important tool for confirming the presence of spinal cord abnormalities in a fetus when a

suspected spinal deformity has been detected on USG. ^{1,6,7} Currently, MRI can be performed without maternal or fetal sedation. We report a new born baby with split cord malformation type 1 with dorsal dermal sinus diagnosed by clinical examination and MRI which clearly shown the lesion.

CASE REPORT

At birth, examination of the newborn showed a small midline swelling in the upper dorsal region, with a tuft of hair on the skin surface (Figure A, B). The swelling was soft and there was no discharge from the lesion. MRI was done on day 2 of the neonatal period showing diastematomyelia in the Lumbosacral region (Figure C), with a thick band separating the two hemicords. This band was seen extending up to the pedicle. There was a sinus track extending from the skin surface up to the spinal canal. There was intraspinal lipoma.

A full term female baby was born by cesarean delivery at the time of resuscitating baby found to have congenital anomaly at the lumbosacral region. Baby cried immediately after birth weighing 2.7 kg have tuft of hair, lipoma and dermal sinus on clinical examination and after MRI confirmed as diastematomyelia type 1 associated with low lying conus, tethered cord and dermal sinus.

Coming to mother obstetric history 2nd sibling the female term baby delivered through normal vaginal delivery weighing 3 kg died at 9 days of life with meningomyelocele. 1st sibling 5 years term female child delivered through normal vaginal delivery weighing 2.5 kg is alive and healthy.

Inspite of 2nd sibling died of congenital anomaly meningomyelocele; the mother didn't have regular antenatal check-ups and not taken folic acid tablets were the main predisposing factor leading to this anomaly.



Figure A: Small midline swelling in the upper dorsal region in lumbosacral region with tuft of hair.



Figure B: Lateral view of baby showing swelling and tuft of hair.

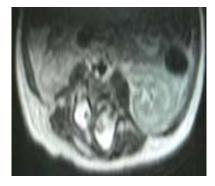


Figure C: MRI showing diastematomyelia.



Figure D: Lumbar tracking, dermal sinus, sinus tracking from surface of skin to dural canal.

DISCUSSION

Diastematomyelia may be an isolated finding or may be associated with other spinal dysraphisms such as myelomeningocele, meningocele, lipoma, neurenteric cyst, and dermal sinus. The vertebral anomalies associated with diastematomyelia include hemivertebra, with kyphosis or scoliosis. There may be associated renal, rectal, and uterine malformations. ¹⁰

The most common location of diastematomyelia is in the thoracolumbar region. Diastematomyelia usually occurs between 9th thoracic and 1st sacral levels of the spinal column with most being at the level of the upper lumbar vertebra .Rarely, it can affect the cervicodorsal region. There is even a case report in the literature of a basicranial diastematomyelia. ¹¹

The extent (or length of spinal cord involved) varies from one affected individual to another. In approximately 60% of patients with diastematomyelia, the two hemicords, each covered by an intact layer of pia arachnoid, travel through a single subarachnoid space surrounded by a single dural sac. Each hemicords has its own anterior spinal artery. This form of diastematomyelia is not accompanied by any bony spur or fibrous band and is rarely symptomatic unless hydromyelia or tethering is present. The other 40% of patients have a bony spur or a fibrous band that passes through the two hemicords. In these cases, the dura and arachnoid are split into two separate dural and arachnoidal sacs, each surrounding the corresponding hemicords which are not necessarily symmetric. Each hemicords contains a central canal, one dorsal horn (giving rise to a dorsal nerve root), and one ventral horn (giving rise to a ventral nerve root.) In some cases the bony spur typically situated at the most inferior aspect of the dural cleft. They advised that if the imaging appears to show otherwise, a second spur (present in about 5% of patients with diastematomyelia) is likely to be present. The conus medullaris is situated below the L2 level in more than 75% of these diastematomyelia patients. Thickening of the filum terminale is seen in over half of the cases. While the level of the cleft is variable, it is most commonly found in the lumbar region. The two

hemicords usually reunite caudally to the cleft. Occasionally, however, the cleft will extend unusually low and the cord will end with two separate coni medullarae and two fila terminale ("Diplomyelia"). 3,4

When the split does not reunite distally to spur, the condition is referred to as a Diplomyelia or true duplication of spinal cord. Females are affected more commonly than males .Split cord malformations are a congenital abnormality and account for ~5% of all congenital spinal defects.

In SCM, there is vertical splitting of the spinal cord. Pang et al. have proposed a classification for SCM.8 According to them, type I SCM consists of two hemicords each contained in a separate dural tube and separated by an osseocartilaginous septum. Type II SCM consists of a single dural sac containing both hemicords; the two hemicords being separated by a nonrigid fibrous septum. Imaging studies rarely show this fibrous septum. This differentiation has surgical importance as type I split cords are technically more difficult to correct and are associated with more surgical morbidity than type II, especially if there is an oblique septum dividing the cord asymmetrically.

The signs and symptoms of diastematomyelia may appear at any time of life, although the diagnosis is usually made in childhood. Cutaneous lesions (or stigmata), such as a hairy patch, dimple, hemangioma, subcutaneous mass, lipoma or teratoma override the affected area of the spine is found in more than half of cases. Neurological symptoms are nonspecific, indistinguishable from other causes of cord tethering. The symptoms are caused by tissue attachments that limit the movement of the spinal cord within the spinal column. These attachments cause an abnormal stretching of the spinal cord. The course of the disorder is progressive. In children, symptoms may include the "stigmata" mentioned above and/or foot and spinal deformities; weakness in the legs; low back pain; scoliosis; and incontinence In adulthood, the signs and symptoms often include progressive sensory and motor problems and loss of bowel and bladder control. This delayed presentation of symptoms is related to the degree of strain placed on the spinal cord over time. Tethered spinal cord syndrome appears to be the result of improper growth of the neural tube during fetal development, and is closely linked to spina bifida.

Tethering may also develop after spinal cord injury and scar tissue can block the flow of fluids around the spinal cord. Fluid pressure may cause cysts to form in the spinal cord, a condition called Syringomyelia .This can lead to additional loss of movement, feeling or the onset of pain or autonomic symptoms.

Cervical diastematomyelia can become symptomatic as a result of acute trauma and can cause major neurological deficits, like hemiparesis, to result from otherwise mild trauma.³

Prenatal diagnosis of this anomaly is usually possible in the early to mid-third trimester. An extra posterior echogenic focus between the fetal spinal laminae was seen with splaying of the posterior elements, thus allowing for early surgical intervention and has a favorable prognosis. Prenate ultrasound could also detect whether the diastematomyelia is isolated, with the skin intact or association with any serious neural tube defects. Progressive neurological lesions may result from the "tethering cord syndrome" (fixation of the spinal cord) by the diastematomyelia phenomenon or any of the associated disorders such as myelodysia, dysraphia of the spinal cord.^{3,4}

Treatment

Surgery

Surgical intervention is warranted in patients who present with new onset neurological signs and symptoms or have a history of progressive neurological manifestations which can be related to this abnormality. The surgical procedure required for the effective treatment of diastematomyelia includes decompression surgery of neural elements and removal of bony spur. This may be accomplished with or without resection and repair of the duplicated dural sacs. Resection and repair of the duplicated dural sacs is preferred since the dural abnormality may partly contribute to the "tethering" process responsible for the symptoms of this condition. 3,4

Asymptomatic patients do not require surgical treatment. These patients should have regular neurological examinations since it is known that the condition can deteriorate. If any progression is identified, then a resection should be performed.

CONCLUSION

Prenatal knowledge of spinal cord anomalies is important for genetic counselling as well as surgical intervention. Since MRI shows these lesions better and with less interobserver variation than USG, in patients with suspected spinal anomalies, either diagnosed on USG or based on clinical and laboratory criteria, fetal MRI should be used prior to further management.

Diastematomyelia can be suspected sonographically as early as the first trimester. If diastematomyelia is found, amniocentesis for AF-AChE and AF-AFP is a reliable indicator of spinal integrity. Intrauterine recognition of diastematomyelia should facilitate appropriate management of the disease, which is important for the prevention of neurological sequelae.

MRI diagnosis in this baby shown type 1 Diastematomyelia associated with low lying conus, tethered cord and dorsal dermal sinus. Presently baby is fine and doing well without any associated neurological deficits. These babies should be followed up at regular

intervals to identify development neurological deficits or associated bowel and bladder dysfunction the baby can develop in near future.

So to prevent neural tube defects and other associated congenital anomalies in newborns; mother should have regular antenatal check-ups, antenatal ultrasound examinations and should take folic acid tablets regularly during gestational period.

Funding: No funding sources Conflict of interest: None declared Ethical approval: Not required

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Cite this article as: Balaji MD, Shivaprakash NC, Thangella PK. Case of congenital diastematomyelia type 1: a rare presentation. Int J Contemp Pediatr 2016;3:273-6.