A tail of sacral agenesis: delayed presentation of meningocele in sacral agenesis

Christopher C. Gillis · Ahmad A. Bader · Michael Boyd

Received: 16 September 2011 / Revised: 28 January 2012 / Accepted: 24 April 2012 / Published online: 8 May 2012
© Springer-Verlag 2012

Abstract

Introduction Sacral agenesis is a congenital condition associated with multiple orthopedic, spinal, abdominal and thoracic organ deformities. Meningocele is commonly found among patients with sacral agenesis.

Description We present the first case in the literature describing a delayed presentation of terminal (posterior) meningocele in an adult patient born with sacral agenesis.

Conclusion Surgical repair was performed and is the best treatment option for significantly large lesions, with post-operative CSF leak being the main complication.

Keywords Spine · Meningocele · Spina bifida · Spinal dysraphism

system [2–6]. Bony defects of the sacrum can have a rare association with terminal myelomeningocele, a rare form of occult spinal dysraphism that may present as a lumbo-sacral mass [7]. Previous case reports of ‘tail-like appendages’ have been seen in pediatric patients with posterior midline protrusions in the lumbosacralcoccygeal region, usually related to occult spinal dysraphism [8]. About half of the cases, as reported, were associated with posterior meningocele or spina bifida occulta [8]. There is no literature reporting a delayed presentation of posterior or terminal meningocele in association with sacral agenesis. This case presents—to our knowledge—the first case of delayed meningocele presentation in an adult patient with sacral agenesis.

Introduction

Sacral agenesis (SA) is an uncommon condition grouped under the caudal regression syndrome. The incidence is reported to be between 0.01 and 0.05 per 1,000 live births [1]. The caudal regression syndrome ranges from a congenital absence of sacral, lumbar and lower thoracic vertebrae to absence of the coccyx, with the majority of abnormalities involving only the sacrum [2]. SA is associated with multiple organ system abnormalities including the genitourinary tract, the hindgut and the respiratory

Case report

History and examination

The patient is a 28-year-old female patient who was born with sacral agenesis (see Fig. 1). The patient had congenital scoliosis with surgical correction and fixation at age five. She also had bowel and bladder dysfunction for which ostomies were done as child. Neurologically, the patient did reasonably well; was able to walk without aids or braces.

At age 20, the patient first noticed a small painless swelling in the lower back between the buttocks. At age 23 she presented to the care of a spinal surgeon when her swelling significantly increased in size. It was causing significant discomfort especially when sitting down. There were also associated headaches on upright posture. There were no new neurological symptoms, pain, or any changes in the pre-existing spinal deformity.

C. C. Gillis (✉) · A. A. Bader · M. Boyd
Division of Neurosurgery, University of British Columbia,
Vancouver, Canada
e-mail: chriscgillis@gmail.com

M. Boyd
Combined Neurosurgical and Orthopedic Spine Program,
University of British Columbia, Vancouver, Canada

Fig. 1 Anterior posterior X-ray of the lumbosacral region showing deficiency of the bony sacrum, fusion of the lower lumbar spine, and an angled pelvis

Fig. 2 A pre-operative photograph demonstrates the large swelling in the lumbosacral region, the size is approximately that of volleyball

Her examination showed marked thoracolumbar kyphoscoliosis. She had a healed midline thoracic scar. An obvious large swelling was seen starting at the mid-lumbar region and going down toward the left buttock (see Fig. 2). It was fluctuant in nature. No skin abnormalities or signs of infection were present. Her gait was abnormal with an equines foot and ankle deformity on the left side. She also

had pelvic asymmetry, with her pelvic tilt higher on the left than the right.

Her neurological examination showed absent ankle reflexes bilaterally. She had 2/5 plantar flexion bilaterally, with 5/5 in all other muscle groups. She had no perianal sensation, and decreased sensation in the S1 distribution.

A spinal magnetic resonance image (MRI) was performed. The MRI demonstrated spinal cord tethering posteriorly to the dura at the level of L5. Syringomyelia was present within the tethered cord at the L2 and L3 level, which was noted to be low lying, extending into the lumbar area. A sacral meningocele was visualized at the level of S1 and extended posteriorly and inferiorly into the posterior subcutaneous tissues just underlying the skin surface. The sac measured approximately 17.9 cm rostral to caudal, 11.3 cm wide and 9.9 cm from anterior to posterior (see Fig. 3). The cauda equina remnant was noted to be scarred and matted posteriorly. Multiple vertebral body fusion abnormalities, as well as hemivertebrae, were present in the lower thoracic and lumbar region. Based on the imaging findings, it was decided that definitive surgery was required to close the defect, resect the meningocele, and de-tether the spinal cord.

Treatment and postoperative course

The patient underwent de-tethering of the spinal cord, resection of the meningocele sac, closure of the dural defect, and closure of the soft tissue defect with bilateral gluteus maximus advancement flaps. The resection, de-tethering and dural closure was performed by the spine surgery service and the soft tissue closure performed by the plastic surgery service. The resected sac was sent for pathological examination. Intraoperative EMG guided the de-tethering as it was performed at the level where nerve stimulation did not elicit any lower limbs muscle contraction. This allowed for the preservation of the patient's functioning nerve roots.

Postoperative recovery was complicated by recurrent CSF leaks. On postoperative day five the patient developed a CSF leak from the closure. A lumbar drain was inserted percutaneously in the OR under fluoroscopic guidance due to the complex bony anatomy of the sacral region and recent surgery. The drain was functioning for 72 h, until it became blocked. The incision was reinforced and repeat fluoroscopic insertion of a lumbar drain attempted but failed due to inability to satisfactorily place the drain.

The patient was then brought to the operating room for a surgical management of her CSF leak. In this repeat procedure occurred 3 weeks after the initial meningocele repair. The original surgical incision was re-opened and a small dural defect identified intraoperatively with Valsalva maneuver adjunct. The small defect was closed using a

Fig. 3 Preoperative magnetic resonance images of the patient. **a** T2 MRI sagittal view showing the large meningocele sac extending posteriorly just under the skin. The *arrow* illustrates the meningocele extending inferior from the dura. The sac measures approximately 17.9 cm rostral to caudal $\times$ 11.3 cm wide $\times$ 9.9 cm anterior to posterior. **b** T2 MRI sagittal view showing syringomyelocele in the

lower spinal cord and showing the multiple vertebral bodies deformities in the lumbar region. **c** T2 MRI axial cuts showing the meningocele starting in the *midline* and then directed toward the left buttock. **d** T2 MRI coronal views demonstrating the “tail”-like appearance of the meningocele extending between the buttocks

running 5-0 prolene stitch. A repeat valsalva after repair confirmed no further CSF leak. Bovine pericardium dural replacement graft was sutured over the dural defect, dural collagen graft matrix was placed in a double layer over the dura, and fibrin sealant was also placed over the closure. After the dural closure another rostral incision was opened and a laminotomy performed to place a lumbar drain under direct vision.

Unfortunately the repeat operation failed after 2 weeks with recurrent CSF leak. This resulted in operative management with a lumboperitoneal shunt. The shunt was placed intradurally through the previous lumbar incision for repair of the meningocele. The dura was identified and opened, providing access to the subarachnoid space where several centimeters of shunt catheter were inserted easily.

The shunt valve and reservoir was placed in the patient's right flank through a separate incision. The valve chosen was a ‘horizontal/vertical’ accommodation valve in efforts to avoid siphoning on upright position.

The lumbar catheter was connected to the reservoir, the reservoir to the shunt valve and the valve to the peritoneal catheter. The abdominal catheter was placed with the assistance of General Surgery under direct surgical vision, given that previous colostomy and urostomy procedures had been performed.

Two weeks after lumboperitoneal shunt insertion, there was recurrent CSF leak now accompanied by signs and symptoms of meningitis. Given the possibility of lumboperitoneal shunt infection repeat surgery was performed. Intraoperatively, purulent discharge was identified leaking

Fig. 4 Follow up Magnetic Resonance images 3 years after the closure surgery. **a** T2 MRI sagittal view shows the absence of the meningocele sac, and satisfactory wound closure with muscle flap reconstruction. **b** T2 MRI showing the presence of the syringomyelia in the distal spinal cord; which was unchanged from the preoperative

images. The *white arrows* demonstrate the plane of closure of the reconstruction. **c**: T2 MRI axial cuts showing the satisfactory healing of the resection defect with muscle flaps, with no residual meningocele sac

back to the valve and the peritoneal catheter was removed. The lumbar catheter of the shunt was externalized as a lumbar drain. The patient was treated with appropriate antibiotics and the drain removed 2 weeks later. Throughout this prolonged hospital stay, she experienced no new neurological deficits.

The patient was followed periodically by the attending surgeon, and had no recurrence of the swelling, no further CSF leak, and no deterioration in her neurological status when last seen at follow-up approximately 8 years after removal of the lumboperitoneal shunt. She had spinal imaging, which confirmed good surgical results, with no residual meningocele sac (see Fig. 4).

The pathology of the resected sac was reported as consistent with meningocele.

Discussion

Neural tube defects (NTD) are the second most common type of birth defect after congenital heart defects, and myelomeningocele is the most common form of neural tube defect, accounting for greater than 90 % of cases of spina bifida. A posterior meningocele, however, represents the least common form of neural tube defect [1, 9]. The incidence of NTD ranges from 1.0 to 10.0 per 1,000 live births divided into two major categories of equal frequency: anencephaly and spina bifida [9]. Meningocele results from a developmental failure in the caudal end of the neural tube, resulting in a sac containing cerebrospinal fluid, meninges, and overlying skin. The development of the spinal cord is normal and there is usually no associated

neurologic deficit, although there is an association with a tethered spinal cord. In the United States approximately 1,500 infants are born with myelomeningocele each year, but the rate of myelomeningocele and other neural tube defects has declined over the last 3 decades [8, 9].

The association of SA with an anterior meningocele is demonstrated clearly in the Currarino Syndrome. Currarino Syndrome is an autosomal dominant disorder (chromosome 7q36) that includes bony sacral defects, an anorectal malformation and a presacral mass. The mass can be a teratoma, an anterior meningocele, an anterior myelomeningocele or a combination of both. These patients usually have constipation with the cause hypothetically related to either anorectal malformations or to the size of the presacral mass [6].

The gene associated with Currarino Syndrome has been identified as HLXB9, a homeodomain-containing transcription factor, encoding for nuclear protein HB9 which, when mutated, is thought to cause a loss of function [10–12]. HLXB9 is a homeobox gene encoding the nuclear protein HB9. Homeobox genes are essential for normal morphogenesis and are known to function in the regulation of human embryonic development [10, 12]. This mutation has been shown in association with SA both in familial and approximately 30 % of sporadic cases [10–12]. SA can also occur within the VACTERL Association [13].

The embryogenesis of SA is unclear. It has been proposed that the malformation results from an excessive physiologic regression of the embryonic tail thus leading to the term ‘caudal regression syndrome’. Catala et al. [13] found that the developmental ‘regression of the embryonic tail’ actually occurs through active incorporation of caudal provertebrae into the sacrum and through fusion of the most caudal vertebrae. This contrasts with the hypothesis of an excessive physiologic regression. The loss of the embryologic tail is actually a result of fusion and reshaping of the caudal elements [13].

The majority of SA cases are sporadic, non-syndromic, non-familial cases. A review of 50 patients with SA by Emami-Naeini et al. [14] demonstrated that none of the included patients had a familial history of SA. Nonsyndromic SA is most commonly associated with the clinical triad of caudal vertebral body agenesis, anorectal, and genitourinary abnormalities [1]. Musculoskeletal abnormalities associated with SA include a shortened intergluteal cleft, flattened buttock, hip dysplasia, hip and knee flexion contractures, distal leg atrophy, talipes and other foot deformities. The patient in this case did demonstrate, along with sacral agenesis, multiple congenital fusion abnormalities of the thoracic and lumbar vertebral bodies leading to a congenital scoliosis, uterus didelphy, renal abnormalities including renal cysts and bilateral hydronephrosis, and previously had required urostomy and colostomy procedures as a child related to lack of bladder and

bowel function [15]. The neurologic deficit associated with SA correlates with the level of the vertebral defect and neuropathologic studies have demonstrated distal cord dysplasia in patients with lumbar or high SA [1, 9].

These abnormalities are most commonly encountered in the pediatric population, rarely in the adult population [7]. On a review of the available literature there were no noted cases of a delayed formation of a posterior meningocele in an adult with SA. The sole adult patient mentioned in the clinical study of 34 patients with SA by Pang [1] was a 42 year old patient sought treatment for a painful abdominal mass which was found to be an anterior meningocele in association with SA and a tethered cord.

The recommended treatment for this condition is surgical resection. The size of the sac and large midline defect after resection played a role in the incidence of postoperative CSF leak in our patient. Multiple operations were needed to control the CSF leak and were eventually successful but with prolonged hospital stay, multidisciplinary involvement, and an episode of meningitis.

From our experience, we feel that insertion of a lumbar drain electively during the first surgery might have prevented the development of postoperative CSF leak. It should be noted that the insertion of a lumbar drain is challenging in these patients given the associated complex bony anatomy. We were successful in inserting one lumbar drain using fluoroscopy in the operating room, but failed the second attempt after the first one blocked. Even with second drain inserted during second surgery under direct vision, CSF leak still occurred 2 weeks later.

Leaving the thecal sac without soft tissue support on closure likely increases the risk for a dural defect and subsequent CSF leak. The plastic surgery service was involved in closure of soft tissue defect of our patient. Gluteus maximus advancement flaps were used for closure which was noted to be challenging given the abnormal anatomy in the region and large resection defect. There was difficulty in attaching the muscle flap to the deep cavity due to concern about the underlying bowel, and this resulted in a residual CSF collection caudal and ventral to the dural sac (see Fig. 5). After the patient failed the third procedure “repair of dural defect and insertion of lumbar drain under direct vision”, plastic surgery opinion was obtained in relation to repeat muscle flap closure. Due to concerns about limiting the patients’ arm range of motion, a repeat muscle flap closure was not pursued.

Despite the postoperative complications, surgery of symptomatic large terminal meningoceles, as in our patient, is the best treatment option. As demonstrated, a postoperative CSF leak is the most likely complication. The use of intraoperative EMG studies helped to preserve the remaining neurological function of the patient during resection with no new neurological deficits.

Fig. 5 Early postoperative MR images demonstrate a defect in muscle closure which may have contributed to post-operative CSF leak. **a** T2 MRI sagittal cuts showing a CSF collection caudal and ventral to the thecal sac as well as anterior to the muscle flaps

(arrows). **b** T2 MRI fat suppression sequence showing the CSF collection ventral to the muscle flaps, and caudal to the thecal sac (arrows)

Conclusion

We present the first case in the literature describing a delayed presentation of terminal meningocele in an adult patient born with sacral agenesis. The differential diagnosis of a sacral mass in patients with sacral agenesis should include the possibility of meningocele, even as an adult. Surgery is the best option for significantly large lesions, with postoperative CSF leak being the main complication.

Conflict of interest None.

References

1. Andrich J, Kalamchi A, MacEwen GD (1979) Sacral agenesis: a clinical evaluation of its management, heredity, and associated anomalies. *Clin Orthop Relat Res* 139:52–57
2. Pang D (1993) Sacral agenesis and caudal spinal cord malformations. *Neurosurgery* 32(5):755–779
3. Lira E (1938) Agenesis sacro-coccigia. *Rev Ortop Traumatol* 7:231–237
4. Sarnat HB, Case ME, Graviss R (1976) Sacral agenesis. Neurologic and neuropathologic features. *Neurology* 26:1124–1129
5. Guille JT, Benevides B, DeAlba CC, Siriram V, Kumar SJ (2002) Lumbosacral agenesis: a new classification correlating spinal deformity and ambulatory potential. *J Bone Joint Surg* 84(1):32–38
6. Emans PJ, van Aalst J, van Heurn ELW, Marcelis C, Kootstra G, Beets-Tan RGH, Vles JSH, Beuls EAM (2006) The Currarino Triad: neurosurgical considerations. *Neurosurgery* 58:924–929
7. Sim KB, Wang KC, Cho BK (1996) Terminal myelocystocele—a case report. *J Korean Med Sci* 11(2):197–202
8. Singh DK, Kumar B, Sinha VD, Bagaria HR (2008) The human tail: rare lesion with occult spinal dysraphism—a case report. *J Pediatr Surg* 43(9):e41–e43
9. Au KS, Ashley-Koch A, Northrup H (2010) Epidemiologic and genetic aspects of spina bifida and other neural tube defects. *Dev Disabil Res Rev* 16(1):6–15
10. Kochling J, Karbasiyan M, Reis A (2001) Spectrum of mutations and genotype-phenotype analysis in Currarino syndrome. *Eur J Hum Genet* 9:599–605
11. Catala M (2002) Genetic control of caudal development. *Clin Genet* 61:89–96
12. Ross AJ, Ruiz-Perez V, Wang Y, Hagan DM, Scherer S, Lynch SA et al (1998) A homeobox gene, *HLXB9*, is the major locus for dominantly inherited sacral agenesis. *Nat Genet* 20:358–361
13. Catala M, Ziller C, Lapointe F, Le Douarin NM (2000) The developmental potentials of the caudalmost part of the neural crest are restricted to melanocytes and glia. *Mech Dev* 95:77–87
14. Emami-Naeini P, Rahbar Z, Nejat F, El Khashab M (2010) Neurologic presentations, imaging, and associated anomalies in 50 patients with sacral agenesis. *Neurosurgery* 67:894–900
15. Fletcher JM, Copeland K, Frederick JA et al (2005) Spinal lesion level in spina bifida: a source of neural and cognitive heterogeneity. *J Neurosurg* 102(3 Suppl):268–279