Neonatal Diagnosis of a Presacral Mass in the Presence of Congenital Anal Stenosis and Partial Sacral Agenesis

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● **The simultaneous presentation of clinically symptomatic anal anomalies and roentgenographically demonstrated sacral dysgenesis should alert the pediatric surgeon to investigate for the presence of a presacral malformation. We report on such a case to illustrate a new radiographic technique that facilitates diagnosis and management of complex congenital malformations. A 1-day-old white boy presented with anal stenosis, a scimitar-shaped sacrum, and large anterior and posterior meningoceles. In addition, a distinct presacral tumor—a teratoma—was identified. These malformations were identified utilizing metrizamide myelography and three-dimensional reconstruction computed tomography (CT) scanning. The meningoceles and a tethered cord were successfully corrected utilizing a posterior approach. A diverting colostomy was performed and subsequently taken down. Two years postoperatively, the patient continues to do well. This case demonstrates that this triad of anomalies (presacral mass, sacral dysgenesis, and anorectal malformation), once considered, can be safely detected with modern radiologic techniques and can be expeditiously corrected during infancy before further deterioration occurs.**

INDEX WORDS: Anterior sacral meningocele; presacral teratoma; anal stenosis.

SYMPTOMATIC ANAL anomalies in the presence of roentgenographically demonstrated sacral dysgenesis should alert the pediatric surgeon to suspect a presacral malformation. Anorectal anomalies in the presence of sacral dysgenesis have been previously associated with either a presacral teratoma[1] or an anterior meningocele.[2] We describe a child in whom both an anterior meningocele and a presacral teratoma were identified in the neonatal period. The initial presentation was anal stenosis with inability to pass meconium. A scimitar sacrum was identified at that time. We report this case to emphasize the importance of the simultaneous presentation of these anomalies and to illustrate the use of three-dimensional computed tomography (CT) reconstruction to facilitate diagnosis and management of complex congenital malformations.

CASE REPORT

An 8-pound, 10-ounce white boy was transferred at 24 hours of age to The Johns Hopkins Hospital because of failure to pass meconium. The mother was 17 years old, had ingested codeine during the first trimester, and had smoked one pack of cigarettes per day throughout her pregnancy. The pregnancy was uncomplicated and terminated in a normal vaginal delivery. On initial presentation, the infant had tight anorectal stenosis. A sickle-shaped sacrum was identified on plain roentgenogram. Specifically, the coccyx, the right half of the sacrum, and the right lamina of lumbar spine-5 were not present. CT scan with a metrizamide myelogram and three-dimensional reconstruction revealed a large anterior and posterior meningocele (Fig 1), in addition to a distinct presacral mass. The spinal cord was tethered. The only other anomaly identified was grade-III, left vesicoureteral reflux and a neurogenic bladder, documented on voiding cystourethrogram. There was no other neurologic deficit.

The large anterior and posterior meningoceles were repaired, and a laminectomy was performed through a posterior approach. Purse-string sutures were placed at the neck of the anterior meningocele in order to isolate the other distinct presacral mass from the thecal space. Ten days postoperatively, the patient developed sepsis and acute colonic dilation. A diverting colostomy was required. He recovered and returned to the hospital 4 weeks after discharge with fever and irritability. He was successfully treated for meningitis. While being seen as an outpatient 30 days later, hydrocephalus was detected, necessitating a ventriculoperitoneal shunt. He recovered from these complications without any sequelae. At age 20 months, the anterior presacral mass was excised through an anterior abdominal approach and identified as a benign presacral teratoma (Fig 2). Progressive Hegar dilations were accomplished for his anal stenosis. At age 22 months, the colostomy was taken down. The patient is doing very well and defecating adequately at age 26 months; however, continence is as yet untested. He was able to walk normally at age 13 months and is developing normally.

A paternal great-uncle of the patient had a meningocele. A paternal cousin, age 11, was subsequently found to have constipation, a scimitar sacrum, and an anterior meningocele. She is currently undergoing further evaluation and treatment.

DISCUSSION

An anterior sacral meningocele is a rare congenital defect associated with partial agenesis of the sacrum. It is characterized by an oval-shaped cyst in the pelvis containing cerebrospinal fluid and is encompassed by an inner arachnoid and outer dural membrane. It may contain neural elements. The sickle-shaped sacrum, or scimitar sacrum, is the most common of the sacral

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Presented at the 20th Meeting of the Canadian Association of Paediatric Surgeons, Ottawa, Ontario, Canada, September 21-24, 1988.

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0022-3468/89/2410-0031$03.00/0

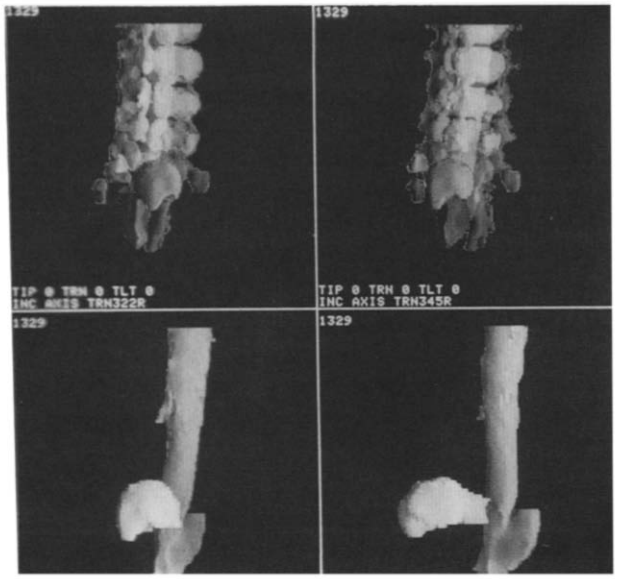

Fig 1. Three-dimensional reconstruction of sacral deformity with metrizamide-filled dural sac revealing anterior meningocele.

anomalies that are invariably present. This lesion, which may remain asymptomatic and is often not identified until adulthood, occurs equally in both genders in childhood.[3] Surgery is recommended in all cases because there is no possibility of spontaneous regression. The complications include partial gastrointestinal obstruction, erosion into the rectum with subsequent meningitis, and pelvic obstruction during labor.[4] A posterior approach with laminectomy and ligation of the communicating stalk, successfully performed in our patient, is recommended since it eliminates any connection between the spinal canal and associated structures.[5]

The genetic predisposition of this lesion has not been ascertained. Cohn and Bay-Nielsen[2] have provided evidence for an x-linked dominant inheritance. Others[6] have provided evidence for an autosomal dominant inheritance, possibly with variable penetrance.[7]

A presacral or sacrococcygeal teratoma originates in the presacral space and contains cellular elements from embryonic ectoderm, endoderm, and mesoderm. The presence of respiratory and squamous epithelium, in addition to glandular tissue (Fig 2), in our patient's tumor is typical of the mature presacral teratoma and aids in its classification as a benign lesion. When a presacral teratoma occurs in association with an anorectal anomaly and sacral dysgenesis, it manifests characteristics that distinguish it from a sacrococcygeal teratoma which presents as a sole anomaly. These differences that occur with other anomalies are: (1) all of these tumors with one exception have been benign,[8] in contrast to an average of a 50% malignancy rate,[9]

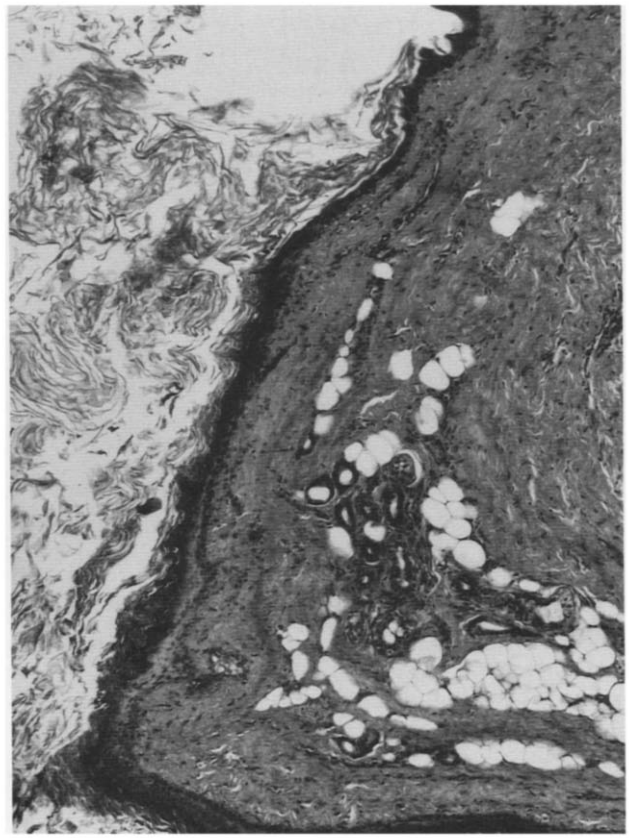

Fig 2. Histology of presacral teratoma. Keratinizing squamous epithelium with sweat glands (H&E, original magnification ×25).

(2) the virtual lack of calcifications,[10] (3) the equal distribution among both genders, (4) familial occurrence,[1] and (5) the presence of sacral deformities.[8]

Our patient had an anterior sacral meningocele and a presacral teratoma documented during infancy. Interestingly, both anomalies: (1) may be familial,[6] (2) most commonly present with constipation, and (3) are associated with sacral deformities.[3,8] Only six other cases in which both presacral malformations were simultaneously present have been reported.[3,11-15]

Undetected neurologic lesions are often present when a spinal dysraphic anomaly occurs simultaneously with anorectal malformation. These lesions include: tethered cord, intradural mass, dural sac stenosis, and diastematomyelia.[16] Early surgical intervention can prevent progressive neurologic deterioration.[17] Our patient had a tethered cord in addition to anterior and posterior meningoceles. All were diagnosed preoperatively and were easily corrected in the initial procedure during infancy.

Three-dimensional spatial relationships of bones and soft tissues are difficult to visualize in standard two-dimensional radiographs, but they are critical to the preoperative planning of the surgeon. Three-

dimensional CT imaging allows for three-dimensional surface reconstruction from contiguous axial CT. In essence, a standard-size CT scan is loaded onto a magnetic tape and processed through a computer program to provide a three-dimensional image. We utilized the CEMAX 1000 (CEMAX, Inc, Santa Clara, CA). This technique has been described previously by Hunter et al[18] for its application in spine disorders. The usefulness of this technique has also been substantiated in craniofacial reconstruction[19] and in the operative planning of procedures for certain orthopedic disorders.[20]

Traditionally, metrizamide myelography with CT scanning has been the standard in the evaluation of spinal dysraphic anomalies. Magnetic resonance imaging (MRI) may be an acceptable alternative; its advantages lie in that it is noninvasive and does not necessitate any ionizing radiation.[21] Recently, Karrer et al[22] suggested the use of high-resolution ultrasonography in search of specific spinal cord lesions in the presence of anorectal malformations and spinal dysraphism. Although this technique remains promising, metrizamide myelography and CT scanning remain the standard. A metrizamide myelogram with a three-dimensional reconstruction of a standard CT scan provides a highly sensitive, specific, and detailed image of anomalies that at the very minimum equals that produced by any other radiographic modality, including MRI and ultrasound. Furthermore, it provides a preoperative view of the anomaly, previously available only under direct intraoperative vision.

Three-dimensional reconstructive CT scanning in pediatric surgery is a promising radiographic technique that warrants further investigation. Our case report demonstrates that a triad of anomalies (presacral mass, sacral dysgenesis, and anorectal malformation) can be safely detected and expeditiously corrected during infancy.

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