Tethered cord syndrome in children with anorectal malformations

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Object. Anorectal malformations are known to be associated with neurological deficits, which may contribute to the disability suffered by patients with these malformations. This study was undertaken to determine the incidence and pattern of sacral abnormalities in children with anorectal malformations, the incidence and nature of the neurological deficits, and the incidence and nature of operable intraspinal abnormalities in patients with this condition.

Methods. Neurological evaluation was performed in 81 children with anorectal malformations. Plain x-ray films were obtained to identify the presence of sacral abnormalities. The patients with neurological deficits were evaluated for the presence of operable intraspinal anomalies, and when such anomalies were identified, correction of the same was undertaken. In 21% of these children radiographic evidence of sacral abnormalities was shown. Fifteen percent of patients harbored neurological deficits, and 10% harbored operable intraspinal anomalies. In addition, one patient had split notochord syndrome. Patients with operable intraspinal anomalies underwent surgical correction, with resultant neurological improvement.

Conclusions. Bone abnormalities of the sacrum, neurological deficits, and operable intraspinal lesions are not uncommon in children with anorectal malformations. Because the neurological deficits can contribute to the disability suffered by these individuals, we recommend routine screening of patients with anorectal malformations and neurological deficits and/or sacral abnormalities for the early identification and treatment of potentially correctable intraspinal lesions.

Key Words • anorectal malformation • imperforate anus • lipomyelomeningocele • sacral agenesis • split notochord syndrome • tethered spinal cord • children

Anorectal malformations are known to be associated with bone deformities of the sacrum. Neurological deficits are known to occur in patients with anorectal malformations, and these deficits were traditionally believed to be caused by developmental dysgenesis of the neural elements. However, recent experience has shown that operable intraspinal lesions are not infrequently seen in children with anorectal anomalies. Because the development of the spinal cord, vertebral column, and anorectal complex is spatially and temporally closely related, it is conceivable that a teratogenic insult occurring at a crucial time can affect all the three systems.

It is important to recognize the association of anorectal malformations with occult dysraphic malformations, because such dysraphic states may complicate the care of these patients. Bladder and bowel symptoms commonly found in this group of patients may be attributed either to the anomaly itself or to surgical procedures, whereas the cause may lie elsewhere.

Abbreviations used in this paper: MR = magnetic resonance; VATER = syndrome of vertebral, anal, tracheoesophageal, renal, and radial anomalies.

Our aim in this article is to emphasize the emerging role for the neurosurgeon and a multidisciplinary team approach in the management of patients with these complex malformations. This study was undertaken to determine: 1) the incidence and pattern of bone abnormalities of the sacrum; 2) the incidence and nature of neurological deficits; and 3) the incidence and nature of operable intraspinal anomalies in children with anorectal malformations.

Clinical Material and Methods

Patient Population

Eighty-one children who attended the Anorectal Clinic at our institution were included in this study. The ages of the patients ranged from newborn to 12 years, and there were 54 boys and 27 girls. There were 46 patients with high, nine with intermediate, and 26 with low anorectal malformations. The Wingspread classification is used in our institution to categorize these children, all of whom underwent a complete neurological evaluation by the neurosurgeon (N.M). Plain x-ray films were obtained in all patients, and we used radiological typing of sacral agenesis according to a system proposed by Pang to classify.
the bone abnormalities of the sacrum seen on these films. In all children in whom imaging of the spinal cord was performed, the imaging studies were obtained after correction of the anorectal malformations was completed. Computerized tomography myelography or MR imaging was performed in children who experienced neurological deficits. Patients in whom there were isolated sacral abnormalities without neurological deficits did not undergo imaging of the spinal cord. In those children in whom operable lesions were encountered, surgical correction was undertaken. Follow up ranged from 6 months to 5 years (mean 18 months).

**Results**

In 17 children (21%) with anorectal malformations, radiographic evidence of bone abnormalities of the sacrum was revealed. Of these, 12 children had high, one had intermediate, and four had low anorectal malformations. Thus, sacral dysgenesis was three times more common in association with high anorectal malformations than low malformations. Among the 17 children with sacral dysgenesis, 10 had Type III agenesis, four had Type IVb, and three had Type IVc dysgenesis. Of the 81 children with anorectal malformations, 12 (15%) experienced neurological deficits (Table 1). In two of the children with neurological deficits no bone abnormalities in the sacrum were demonstrated. The neurological deficits varied from mild distal sensorimotor deficits and limb length discrepancies (Fig. 1) to urinary and bowel incontinence.

The pattern of neurological deficits was no different from that seen in patients with occult spinal dysraphism. In addition to anorectal and neurological anomalies, many patients had orthopedic and genitourinary deformities. The most common orthopedic anomaly was congenital talipes equinovarus, and the most common urological anomaly was renal ectopia. Among the children with neurological deficits, examinations revealed operable intraspinal anomalies in eight: tethered spinal cord in six (Fig. 2), lipomyelomeningocele in one, and tethered spinal cord with transitional lipoma in one. The incidence of intraspinal anomalies was equally distributed between high and low anorectal malformations. In addition to the aforementioned abnormalities, there was one patient with split notochord syndrome accompanied by anal agenesis and dorsal enteric fistula. This child had a thoracic myelomeningocele, weakness of one lower extremity, absent anus, posteriorly displaced scrotum, and a dorsal enteric fistula in the lower back. Plain x-ray films obtained in this child revealed the typical inverted Y split of the lower spine characteristic of split notochord syndrome (Fig. 3). All children with operable intraspinal anomalies underwent surgical correction of their lesions. The nature of improvement following surgical intervention is summarized in Table 1. There was a significant improvement in urinary and sensory symptoms, whereas motor symptoms took longer to resolve.

**TABLE 1**

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age (yrs), Sex</th>
<th>Type of Neurological Deficit</th>
<th>Type of Anorectal Anomaly</th>
<th>Imaging Findings</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>3, M</td>
<td>limb length discrepancy, urinary incontinence</td>
<td>high tethered spinal cord</td>
<td>incontinence improved</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>9, F</td>
<td>urinary &amp; bowel incontinence, sacral sensory loss</td>
<td>low tethered spinal cord w/ transitional lipoma</td>
<td>urinary incontinence improved, no change in bowel incontinence</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>7, F</td>
<td>urinary incontinence, foot deformity, reflex changes</td>
<td>low tethered spinal cord</td>
<td>incontinence improved</td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>9, M</td>
<td>limb length discrepancy, foot deformities, urinary incontinence</td>
<td>high tethered spinal cord</td>
<td>incontinence improved</td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>3, M</td>
<td>foot deformity, wasting of calf muscles, limb length discrepancy</td>
<td>high normal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>12, F</td>
<td>limb length discrepancy, urinary incontinence</td>
<td>low tethered spinal cord</td>
<td>incontinence improved</td>
<td></td>
</tr>
<tr>
<td>7</td>
<td>5, M</td>
<td>limb length discrepancy, mild weakness of 1 lower limb, urinary incontinence</td>
<td>high lipomyelomeningocele</td>
<td>incontinence improved</td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>4, F</td>
<td>urinary incontinence</td>
<td>low normal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>4, M</td>
<td>urinary incontinence</td>
<td>low tethered spinal cord</td>
<td>incontinence improved</td>
<td></td>
</tr>
<tr>
<td>10</td>
<td>6, F</td>
<td>urinary incontinence</td>
<td>high normal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>4, M</td>
<td>urinary incontinence</td>
<td>low normal</td>
<td></td>
<td></td>
</tr>
<tr>
<td>12</td>
<td>5, F</td>
<td>urinary incontinence, distal sensorimotor deficit</td>
<td>high tethered spinal cord</td>
<td>incontinence improved</td>
<td></td>
</tr>
</tbody>
</table>

**Fig. 1.** Photograph showing a child with anorectal malformation, limb length discrepancy, and wasting of the muscles of the lower extremities on one side.
Discussion

Association of Anorectal Malformations and Spinal Dysraphism

Embryogenesis. During development, the precursors of the anorectal, urogenital, and caudal spinal structures are closely related both in space and time. The anorectal and urogenital structures arise from the cloaca, which is present by the 4th week of gestation. Subsequently, the cloaca becomes divided by the urorectal septum into the dorsal hindgut and ventral urogenital sinus. The cloacal membrane forms the anal and urogenital membranes when the urorectal septum fuses with it at the 7th week of gestation. Different types of anorectal and other urogenital anomalies can occur due to abnormal development of the cloaca. During the 4th to 7th week of gestation, the caudal cell mass undergoes differentiation into the caudal neural tube dorsal to the cloaca, with the notochord interposed. Subsequently, the caudal neural tube fuses with the rostral tube formed by neurulation. Retrogressive differentiation of the caudal neural tube leads to the formation of the filum terminale. Because the development of the caudal neural tube and the anorectal complex are closely related in space and time, it is conceivable that a teratogenic insult that occurs during this phase can affect both systems simultaneously, albeit to different extents. It has also been suggested that the notochord influences the development of not only the neural tube but also other adjacent tissues, notably the hindgut. Therefore, abnormal development of the notochord not only produces vertebral anomalies but may also inhibit the formation of the hindgut, thus producing an imperforate anus.

Clinical Studies. The aforementioned association has been highlighted by the work of Carey, et al., who have described a group of concurrent anomalies as the “OEIS” complex (omphalocele, exstrophy, imperforate anus, and spinal defects). Four of their six patients had skin-covered lumbar or sacral meningoceles. Duhamel pointed out that patients with anorectal malformations have a regional defect resulting from an anomalous organization of the caudal pole of the embryo, which he termed caudal regression syndrome. According to this author, the caudal eminence forms the pluripotential source for most of the tissues of the caudal embryo, especially the caudal spinal cord, the anorectal complex, and the genitourinary tract; a teratogenic insult to this structure can lead to abnormalities that involve all three systems.

Anorectal Malformations and Sacral Abnormalities

In our series, there was a 21% incidence of sacral abnormalities. The incidence of these abnormalities in patients with imperforate anus has been reported to vary between 13% and 54%. The high incidence of vertebral anomalies in patients with imperforate anus is well known. These vertebral anomalies vary from hemivertebra and lumbar block vertebra to partial or total sacral agenesis. The association of vertebral anomalies with anorectal malformations was highlighted by Quan and Smith when they described the VATER syndrome, a nonrandom association of vertebral, anal, tracheoesophageal, renal, and radial anomalies. These authors pointed out that on finding one of the VATER anomalies the physician should be alerted to the possibility of the presence of other anomalies associated with this syndrome. Pang has shown that abnormalities of the sacrum can be divided into two types: high sacral malformations with no vertebrae below S-1 and low sacral malformations with preserved S-2 or lower sacral pieces; and low sacral malformations are more likely to be associated with operable intraspinal anomalies. In our present study of children with anorectal malformations there was no patient with a high sacral malformation.

Anorectal Malformations and Intraspinal Anomalies

In our study, 10% of patients with anorectal malformations had operable intraspinal anomalies. In other reported series, the incidence varies from 8 to 36%. The types of intraspinal lesions found in our series are similar to those in other reported series. In other series with a higher incidence of intraspinal abnormalities in anorectal malformations, even neurologically asymptomatic children underwent MR imaging evaluation. We believe that if all our patients (including asymptomatic patients) had undergone examination with MR imaging, the incidence of tethering lesions would have been higher. The incidence of associated neurological abnormalities in our and other reported series is small but significant. In a recent study, Long, et al. evaluated 86 patients with anorectal malformations by using plain radiography and MR imaging. Magnetic resonance imaging revealed that 36%...
Tethered cord and anorectal malformations

of these children had occult spinal dysraphic lesions, including some children in whom normal results had been demonstrated on plain x-ray films. Appignani and colleagues evaluated 92 patients with imperforate anus, cloacal malformation, and cloacal extrophy and found evidence of myelodysplasia in 34% of patients with high imperforate anus, 46% in patients with cloacal malformations, and 100% in patients with cloacal extrophy. Thus, in their series, the incidence of associated occult dysraphic malformations increased in direct proportion to the severity of the anorectal malformations. Heij, et al.,10 reported a 30% and 50% incidence of intraspinal anomalies in children with low and high anorectal malformations, respectively. However, in our series the incidence of intraspinal anomalies was equal in patients with both high and low anorectal malformations. Tsakayannis and Schamberger21 reported the association of imperforate anus with occult spinal dysraphism. They found that 14% of children with imperforate anus harbored associated occult spinal dysraphic lesions. They recommended routine screening of children with imperforate anus by using ultrasonography or MR studies, and they advocated the early neurosurgical correction of these lesions.

Cohen evaluated five infants who harbored cloacal extrophy and found four who harbored terminal myelocystoceles and one who harbored lipomeningocele. He postulated that with eversion of the bladder and bowel through the defect in the anterior abdominal wall, the notochord will be displaced anteriorly. Because the notochordal influence on neurulation as well as retrogressive differentiation of the caudal neural tube is dependent on the distance between the notochord and the developing neural tube, such an anterior displacement of the notochord results in impaired retrogressive differentiation of the caudal neural tube resulting in occult spinal dysraphism.

Screening of Patients With Anorectal Malformations

Magnetic resonance imaging is the modality of choice for screening patients with anorectal malformations for the presence of spinal dysraphic lesions. In a recent study, Heij, et al.,10 performed MR imaging evaluation in 43 patients with anorectal malformations and found abnormalities of the spinal cord and spine in 46.5% of these patients. They therefore recommended routine in MR imaging screening of patients with anorectal malformations. In addition, as shown by Pringle, et al.,18 it may also be a useful adjunct in planning pull-through procedures, because MR imaging clearly reveals the status of the pelvic muscle complex. Genitourinary anomalies, which are commonly associated with anorectal malformations, can also be studied. Beek, et al.,3 have studied the value are commonly associated with anorectal malformations, striated muscle complex. Genitourinary anomalies, which

FIG. 3. Plain x-ray film, anteroposterior view, obtained in a child with split notochord syndrome, demonstrating the typical inverted Y split of the spine.

lineating the nature of the deformity, if results on plain x-ray films and ultrasound were normal, MR imaging examination in such children would be superfluous. In our institution, spinal sonography is currently being used as a screening tool for infants with suspected occult spinal dysraphism. However, none of the children in the present study underwent spinal sonography.

Response to Treatment

Previously it was believed that neurological deficits in children with imperforate anus were caused by a simple deficiency of the spinal nerve roots rather than by spinal cord tethering. Moreover, urinary and/or bowel incontinence that develops in patients with anorectal malformations may also be incorrectly attributed to the pull-through procedures. However, as shown by this and other studies, it is not uncommon to find neurological deficits in children with anorectal malformations, in whom these associated deficits may contribute significantly to their disability.

It is well known that well-established neurological deficits in children with tethered spinal cords do not always resolve completely, especially if they have been present for a long time. Our results are in partial agreement with this view. However, unlike conventional neurosurgical wisdom, which states that among all the neurological deficits, sphincters are more vulnerable and less likely to recover after treatment, our experience with this and other groups of patients with occult spinal dysraphic lesions (unpublished data) shows that patients with these lesions.
have a better chance of recovering sphincter functions, even if the deficits have been present for a long time. This might be because these sphincter disturbances are due to the metabolic disturbances in the sacral cord rather than irreversible damage. However, because these deficits do not always completely resolve, it is imperative that the pediatric and neurosurgical teams treating these patients take a proactive role in identifying such correctable lesions, and, where applicable, a preemptive untethering procedure should be performed.

Conclusions
Among the children with anorectal malformations in our study, 21% had bone abnormalities of the sacrum, 15% had neurological deficits, and 10% had operable intraspinal anomalies. Thus, a small but significant percentage of patients with anorectal malformations may harbor occult spinal dysraphic lesions. Because these lesions and the resultant neurological deficits may contribute significantly to the disability suffered by these patients, early diagnosis is mandatory. Therefore, we believe that all children with anorectal malformations and neurological deficits and/or sacral abnormalities should be carefully screened for the early identification and correction of operable intraspinal anomalies.

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References

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