Introduction

Anorectal malformations (ARMs) include a relatively complex set of congenital malformations. Some patients with ARMs suffer late onset of neurovesical dysfunction and/or orthopedic symptoms, which may include febrile and urinary incontinence, lower extremity weakness, numbness, changes in muscle tone, and back and leg pain. The association between spinal cord anomalies and imperforate anus has been well recognized. Until now, the incidence of tethered cord has been assumed to be more significant in patients with a high lesion of the imperforate anus. However, recent reports suggest that the incidence of tethered cord in patients with low lesions is no lower than that in those with high lesions.

Purpose

To review the incidence of spinal cord anomalies and to compare them between imperforate anus patients with low and high lesions, including intermediate lesions.

Materials and Methods

During the period from March 2002 to October 2006, 50 post operatively patients with imperforate anus underwent lumbar magnetic resonance imaging (MRI) at our institution.

Males, 30 cases; females, 20 cases.

Mean age of 9 years at the time of the lumbar MRI (range, 11 days to 25 years old).

Low lesions, 11 cases; 39 cases with intermediate and high lesions (all defined as high lesions for the purpose of this study).

Anatomical reconstruction of imperforate anus was successful in all 50 cases.

MRI technique

- MRI machine
  - Intera 1.5T or 1.0T (Philips)
- MRI protocol (Table)
  - Slice thickness, 3-5mm
- General anesthesia or sedation for neonates, infants and young children.

We retrospectively reviewed the following factors:
1. The incidence and description of symptoms for both the low and high lesions.
2. The incidence and findings of spinal cord anomalies between the low and high lesions.
3. The presence of vertebral anomalies between the low and high lesions.

Results

Results 1-1 Incidence of symptoms (Figure 1)

- The overall incidence of symptoms was 31 of 50 (62%) cases.
- Symptoms were present in 7 of 11 (64%) cases with low lesions and in 24 of 39 (62%) cases with high lesions.

Results 1-2 Description of symptoms (Figure 2)

- The symptoms included urinary and fecal incontinence, lower extremity numbness and leg pain.
- Urinary and fecal incontinence were the most frequent symptoms in both the low and high lesion groups. Both of urinary/fecal and lower extremity symptoms were present in 2 of 7 cases with low lesions and in 2 of 24 cases with high lesions.

Results 2-1 Incidence of spinal cord anomalies (Figure 3)

- Eight of 11 (73%) cases with low lesions had spinal cord anomalies, as did 22 of 39 (56%) cases with high lesions.
- The incidence of spinal cord anomalies did not differ statistically between cases with low and high lesions (2x2 cross tabs and Fisher’s exact test, P>0.05).

Results 2-2 Findings of spinal cord anomalies (Figure 4)

- MR images showed spinal lipoma (lipomyelomenigocele, intradural lipoma, fibrolipoma of the filum terminale), anterior sacral meningocele, and caudal regression syndrome. The other findings showed tethered cord without spinal lipoma and unrecognized syringomyelia.

- Of the above anomalies, spinal lipoma was most frequent in both groups. Anterior sacral meningocele and caudal regression syndrome were identified in 1 case with low lesions and 1 case with high lesions.

Results 2-3 Incidence of tethered cord (Figure 5)

- Tethered cord was revealed in 4 of 11 (36%) cases with low lesions and in 17 of 39 (44%) cases with high lesions.
- Among the 21 cases with tethered cord, 4 (1 case with low lesions and 3 cases with high lesions) also showed syringomyelia.

Results 3 Presence of vertebral anomalies (Figure 6, 7)

- Vertebral anomalies were found in 4 (36%) cases with low lesions and in 26 (67%) cases with high lesions.
- The vertebral anomalies found included sacral hypogogenesis, hemivertebra, fusion, spina bifida and absence of the coccyx.
- Four of 11 (36%) cases with low lesions and 19 of 39 (49%) cases with high lesions had both vertebral and spinal cord anomalies. On the other hand, 4 (36%) cases with low lesions and 3 (8%) cases with high lesions had spinal cord anomalies without vertebral anomalies.
Discussion

ARMS are frequently associated with other anomalies, especially of the spinal cord, spine and urogenital system. The overall incidence of patients having one or more associated anomalies was 45.2%.

Incidence and description of symptoms

The symptoms found in conjunction with imperforate anus include fecal incontinence, neurovesical dysfunction, gait disturbance and spastic abnormalities of the lower extremities. The onset of symptoms is usually occurs in childhood and may be slowly progressive. Neurovesical dysfunction is primarily related to tethered cord or to an iatrogenic nerve injury. Once symptoms appear, they are often irreversible despite surgical intervention and may have a significant impact on the continence of patients with imperforate anus. Surgical release of a tethered cord before the onset of neurological problems may prevent the onset of these symptoms.

Incidence and findings of spinal cord anomalies

Heij et al. have demonstrated that the incidence of tethered cord is higher in patients with high lesions (50%) than in patients with low lesions (30%). However, Golonka et al. report that the incidence of tethered cord in children with low lesions is no lower than that in those with high lesions.

In the present study, patients with spinal cord anomalies accounted for 73% of those with low lesions and for and for 56% of those with high lesions, tethered cord was identified in 36% of low lesion patients and 44% of the high lesion group. These results are similar to those reported in previous studies.

Presence of vertebral anomalies

Vertebral anomalies have been reported to be twice as common in patients with high lesions than in those patients with low lesions. On the other hand, spinal cord anomalies without vertebral anomalies were frequently found in patients with low lesions in the present study. Some authors recommend MRI evaluation of all patients with ARMS because spinal cord anomalies, including tethered cord, are known to occur in patients without sacral anomalies as well as in those with low ARM.

A number of authors have recommended ultrasound screening of infants with imperforate anus followed by MRI to confirm abnormal findings. Ultrasound is an ideal screening tool for infants less than 3 months of age. Mosiello et al. recommend that all patients be screened with MRI at age 6 to 12 months. MRI is the most sensitive modality for detecting spinal cord anomalies as well as for detecting vertebral anomalies.

Conclusion

Owing to the high incidence of spinal cord anomalies in patients with imperforate anus, MRI is the best type of imaging study to detect such anomalies regardless of the type of imperforate anus.