

Occult Spinal Dysraphism in Children with Anorectal Malformation

MWY LEUNG, BPY WONG, TW FAN, NSY CHAO, KW CHUNG, WK KWOK, KKW LIU

Abstract

Purpose: To review the association of occult spinal dysraphism in children with anorectal malformation. **Methods:** Fifty-eight children with anorectal malformation were reviewed. Twenty patients had high anomalies, whereas 6 had intermediate and 32 had low anomalies. Magnetic resonance imaging (MRI) of lumbosacral spine was performed on 10 children, because of abnormal preliminary ultrasonographic findings or persistent symptoms after surgery. **Results:** Four children had positive findings from the MRI. Tethered cord and intraspinal lipoma are the most common abnormalities. Lumbosacral spine X-ray was abnormal in only one patient. All patients had urodynamic assessment and abnormal cystometric finding was evident in two patients. **Conclusions:** Occult spinal dysraphism is common in children with anorectal malformation. A normal lumbosacral spine X-ray does not exclude the diagnosis. MRI of spine should be performed in suspicious cases. Urodynamic assessment is necessary to identify associated neurovesical dysfunction.

Key words Imperforate anus; Spinal dysraphism

Introduction

Anorectal malformation (ARM) is a common congenital anomaly with an incidence of 1 in 4000 to 5000 live births. According to the severity, it can be

classified as low, intermediate and high anomalies.¹ ARM is commonly associated with oesophageal atresia, cardiac, urological, vertebral and upper limb anomalies (VACTERL association).² Sacral agenesis, hemivertebra, bifid vertebral bodies, extra or absent ribs are common vertebral anomalies. However, intraspinal abnormalities such as tethered cord and intraspinal lipoma have been reported to be associated with ARM without any skeletal anomaly.³ Occult spinal dysraphism (OSD) may account for the abnormal gait, voiding dysfunction and fecal incontinence of the patients after surgery. This article reviews the children who have OSD in association with ARM in our centre.

Division of Paediatric Surgery, Department of Surgery, Queen Elizabeth Hospital, 30 Gascoigne Road, Kowloon, Hong Kong, China

MWY LEUNG (梁偉業) FHKAM(Surg)
NSY CHAO (趙式言) FHKAM(Surg)
KW CHUNG (鍾廣槐) FHKAM(Surg)
WK KWOK (郭永堅) FHKAM(Surg)

Division of Paediatric Surgery, Department of Surgery, United Christian Hospital, 130 Hip Wo Street, Kwun Tong, Kowloon, Hong Kong, China

BPY WONG (王珮瑤) FHKAM(Surg)
KKW LIU (廖鑑榮) FHKAM(Surg)

Department of Radiology & Imaging, Queen Elizabeth Hospital, 30 Gascoigne Road, Kowloon, Hong Kong, China

TW FAN (范子和) FHKAM(Radiol)

Correspondence to: Dr MWY LEUNG

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Methods

Fifty-eight children, 34 boys and 24 girls, with anorectal malformation were managed in our centre from 1995 to 2004. According to the Wingspread classification,¹ 20 children had high anomaly, 6 had intermediate and 32 had low anomaly. Cut back or limited posterior sagittal anorectoplasty (PSARP) were performed for patients with low ARM. PSARP was performed in patients with

intermediate anomalies. PSARP or abdominoperineal pullthrough operation (laparoscopic or open) was performed for high anomalies. Post-operatively, the patients were followed up in our colorectal continence clinic. Three patients (5.2%) had post-operative anal stenosis and 8 patients (13.8%) had anal mucosal prolapse. In our centre, the bowel function of patients was assessed at the age of three years. Median age of patient was 5 years at the time of first bowel continence assessment.

A multi-disciplinary management programme involves oral medications, enemas and biofeedback pelvic floor muscle training. Fifty-three patients (91.3%) had complete social continence (less than one episode or soiling per month) after bowel management program. All but 3 patients are still having regular follow up in our bowel management clinic with a default rate of 5.2%.

Ten patients (17.2%) had vertebral anomalies shown in plain X-ray. Nine patients had hemivertebra of thoracolumbar region and absent ribs but no abnormality of the sacral spine. They all have complete social continence. One patient had no segmentation of sacrum and subsequently found to have OSD. Six patients had ultrasonogram (USG) of lumbosacral spine done in neonatal period with one showing low-lying conus of spinal cord. Pelvic and sacral magnetic resonance imaging (MRI) is not a routine investigation in our centre. The indication for MRI spine is either an abnormal USG or patients with orthopaedic symptoms or persistent fecal with/without urinary incontinence after bowel management programme.

One infant had abnormal USG finding and 9 children had persistent fecal incontinence or associated lower urinary tract symptoms. These ten children had pelvic and spinal MRI performed.

Results

Four children, aged 3 months to 17 years, had abnormal lumbosacral MRI findings. Intraspinous lipoma (3), low lying conus medullaris (3), thickened filum terminale (2) and

syringomyelia (1) were evident. Two patients had high type anorectal malformations and two had low anomalies (Table 1). Plain X-ray of lumbosacral spine was performed in all 4 patients with only one patient showing abnormal finding of absent segmentation of sacrum. One of the 4 patients had previous USG spine performed in neonatal period, showing low lying conus which correlated with the MRI findings. Owing to the abnormal MRI findings in these patients, urodynamic study was performed. Two patients had abnormal cystometric findings. One infant had hyper-reflexic bladder with detrusor muscle overactivity. One 7-year boy had detrusor overactivity, detrusor-sphincter dyssynergia, high voiding pressure with residual urine, bilateral vesicoureteric reflux and renal scarring (Figure 1). Clean intermittent catheterization and endoscopic subtrigonal injection of dextranomer in sodium hyaluronan (Deflux™) was performed. There was no orthopaedic symptom in all patients. All patients were referred to paediatric neurosurgeon for assessment. Untethering of spinal cord was performed in two patients.

Discussion

Occult spinal dysraphism is associated with anorectal malformation. It is reported that spinal cord abnormalities occurred in 9% to 57% of children with anorectal malformation.^{4,5} Tethered spinal cord is a developmental abnormality, in which the lumbosacral spinal cord is prevented from ascent in the spinal canal. It is associated with thickened filum, intraspinal lipoma and syringomyelia. The association of anorectal malformation and spinal dysraphism is demonstrated by the split notochord model in fetal rats.⁶ It is generally believed that the prevalence of OSD is higher in high anorectal anomalies (e.g. rectovesical fistula) than in low anomalies (e.g. ectopic anus).⁷ However, some literatures report equal or even higher prevalence of OSD in low anorectal anomalies.⁸

It is still controversial what routine spinal radiological

Table 1 Abnormal magnetic resonance imaging findings in children with anorectal malformation

Age of patients	Anorectal malformation type	Intraspinous lipoma	Low lying conus	Thickened filum	Syringomyelia
6 months	Low	✓	✓	–	✓
7 years	High	✓	✓	✓	–
9 years	Low	–	✓	–	–
17 years	High	✓	–	✓	–

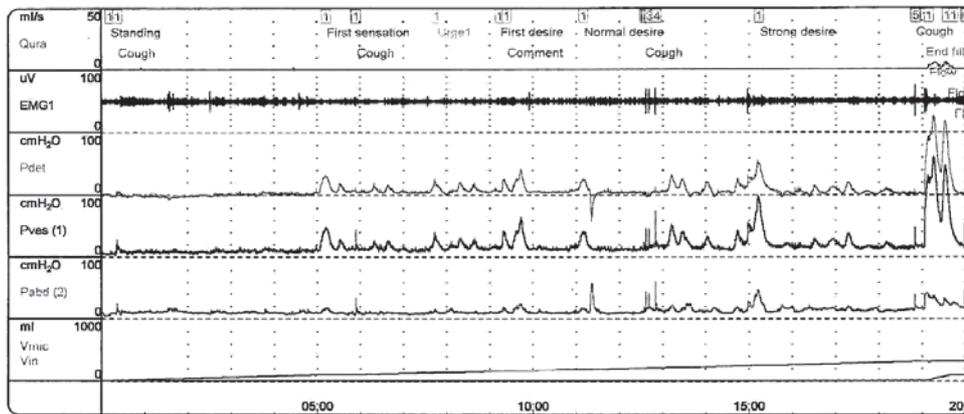


Figure 1 Detrusor overactivity, detrusor-sphincter dyssynergia and high voiding pressure in a child with occult spinal dysraphism.

imaging should be performed in newly diagnosed neonates with ARM.⁹ Plain X-ray is not a sensitive investigation to exclude spinal dysraphism. In our series of 4 OSD patients, only one had abnormal sacral skeletal abnormality. USG of lumbosacral spine is a non-invasive and sensitive test in neonates. The bony or cartilaginous malformations and the level of conus can be detected. We performed USG of spine in 6 patients in the neonatal period with one positive finding of low lying cord. However the sensitivity of USG of spine to detect OSD decreases beyond neonatal period because of ossification of sacral bone. We recommend that in every newborn with ARM, USG of spine should now be performed as routine together with USG of urinary system. MRI spine is indicated if the USG of spine is abnormal (Figure 2).

It is debatable whether MRI of spine should be performed in all newly diagnosed neonates with ARM. The study requires deep sedation or general anesthesia of the neonates. Also the cost-effectiveness of MRI should be considered.¹⁰ It is also controversial to diagnose and treat all OSD patients without symptoms. We believe that MRI of lumbosacral spine should be performed if the USG spine is abnormal, or if the patients have persistent bowel incontinence, urological or orthopaedic symptoms after anorectoplasty. In our series, 10 patients with either abnormal USG or persistent incontinence after bowel management program had MRI of spine performed, and 4 had positive findings of OSD (Figure 3).

In children with ARM and OSD, there is higher incidence of neurovesical dysfunction.¹¹ Urodynamic abnormalities, including detrusor overactivity, detrusor-sphincter dyssynergia, distended bladder and lazy bladder are reported in 32% to 57% of patients.⁵ Urinary incontinence, urinary tract infection, vesicoureteric reflux and renal

scarring can occur. Fifty percent of our patients with ARM and OSD had abnormal urodynamic findings. One boy had detrusor overactivity and high voiding pressure. Clean intermittent catheterization and endoscopic subtrigonal injection of dextranomer in sodium hyaluronan (DefluxTM) was performed to prevent further renal scarring and deterioration in renal function. Thus, we recommend routine urodynamic study in children with ARM and OSD, and long term follow up is required in children with neurovesical problems.

Bademci et al reported that the prevalence of primary tethered cord syndrome in the normal population is 0.1%.¹² In our series, the prevalence of OSD is 4/58 or 6.9%. It is still controversial whether prophylactic untethering of spinal cord is beneficial to children with ARM and OSD. Some authors suggest that prophylactic untethering of cord should be considered to prevent bowel and urinary symptoms in future.^{4,13} However, we have seen patients remaining asymptomatic with conservative management. Delayed



Figure 2 Ultrasound scan of lumbar spine shows a linear hyperechoic structure compatible with fatty filum.



Figure 3 MRI Sagittal T1 weighted image of the spine, a hyperintense linear structure compatible with fatty filum is seen. The spinal cord is not low lying in this patient.

surgery in patients with bowel and urinary problems can arrest or sometimes improve the symptoms.¹⁴ Untethering of cord is not without complications. Post-operative meningitis, CSF fistula, pseudomeningocele have been reported. Meyrat et al reported the postoperative re-tethering rate was as high as 15%.¹⁵ Nevertheless, we think that early referral of children with ARM with OSD to a paediatric neurosurgeon is necessary for assessment.

In conclusion, occult spinal dysraphism is associated with anorectal malformation. Routine USG of lumbosacral spine is recommended in neonatal period. MRI spine is indicated in selected patients with an abnormal USG of spine, urological/orthopaedic symptoms or persistent incontinence not responsive to bowel management.

Urodynamic study and early neurosurgical referral are indicated in patients who are symptomatic or have abnormal MRI findings.

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