Pediatric Surgery International (2019) 35:221–226
https://doi.org/10.1007/s00383-018-4399-x

ORIGINAL ARTICLE

Tethered cord in children with anorectal malformations with emphasis on rectobladder neck fistula

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Accepted: 18 October 2018 / Published online: 9 November 2018

Abstract

Purpose To find the incidence of tethered cord (TC) in patients with anorectal malformations (ARM) and to determine the relationship between bowel/urinary control and TC in a subset of patients with rectobladder neck fistula (RBNF).

Methods The database of a tertiary medical center was retrospectively collected for all patients treated for ARM from 1980 to 2012. All patients with TC and RBNF were identified.

Results Among 790 patients, who underwent screening for TC, 285(36%) were diagnosed with TC. Eleven of 37 screened patients with RBNF were diagnosed with TC. The median follow-up period was 49 months (range 2–222 months). TC was diagnosed in 3/18(16.6%) patients with sacral ratio (SR) ≥ 0.7; 4/12(33.3%) with SR 0.41–0.69; and 4/7(57.1%) patients with SR 0–0.4. The association of TC in RBNF patients had a negative influence in the prognosis for bowel and urinary control.

Conclusion The incidence of TC among patients with ARM is 36%. Incidence of TC among patients with RBNF correlates with SR value and is higher with lower SR. Patients with RBNF and TC have dismal prognosis for bowel control, unrelated to their SR status. Many unresolved questions related to the management of ARM patients with asymptomatic TC still remain.

Keywords Anorectal malformations · Imperforate anus · Tethered cord · Rectobladder neck fistula

Introduction

Tethered cord (TC) is a morphologic term [1], coined by Hoffman et al. in 1976 [2], describing the radiographic picture of the fixation of the filum terminale, which normally terminates above L-2 endplate [3], resulting in limitation of the caudal-cephalic ascent of the conus medullaris within the spinal canal [4, 5].

It was suggested that development and growth of the vertebral column may progressively stretch the tethered spinal cord and might be the source of neurologic deterioration [6], manifested as the tethered cord syndrome (TCS). Clinical manifestations of TCS may include gait disturbances, anatomic deformities, lower extremity weakness, pain, motor and sensory deficits and bowel/bladder dysfunctions [2, 5, 7, 8].

In recent years, there has been a rising trend in the diagnosis of tethered cord owing to the increased use of imaging studies and increased recognition of the condition [9]. It is generally accepted that surgical intervention is indicated in patients with progression or new onset of symptoms [6, 8]. Surgically easing the tension on the cord leads to improvement or stabilization of motor/sensory function in most patients. According to some investigators, the earlier the intervention after symptom development, the better the recovery [8]. However, the reported rates of improvement in bowel and bladder function are less convincing [10].

It is well recognized that anorectal malformations (ARM_S) may be associated with spinal cord tethering as part of the spectrum of occult spinal dysraphism [11–13]. As the development of the caudal neural tube and anorectal complex are closely related in time and space, an insult to the primitive caudal cell mass during this phase may affect both systems to various extents [12, 14].

The reported incidence of TC among patients with ARM_S varies considerably in different studies and ranges from 9 to 52% [1, 4, 12, 15–17] and screening for TC in newborns

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with ARM_S has become standard practice [4, 11, 12, 18]. The higher volume of imaging studies has increased the detection rates of tethered cord, but it has also raised important questions regarding its management in asymptomatic cases, the benefit of prophylactic untethering, and the significance of isolated tethered cord in terms of functional prognosis. It is also unclear, whether TC has the same natural history in the setting of ARM_S as in other spinal dysraphism [1, 8].

The aim of the present study was to review three decades of experience of a pioneering group of experts in ARMs with tethered cord. We sought to determine the prevalence of tethered cord in patients with ARMs with a specific focus on rectobladder neck fistula and to evaluate the relationship between bowel and bladder control, and the radiologic finding of tethered cord in this subset of patients.

Materials and methods

The database of a tertiary medical center was retrospectively reviewed for all patients treated for ARM from December 1980 to September 2012, who underwent screening for tethered cord. The types of ARM and the screening results were recorded. Patients with rectobladder neck fistula were identified for further analysis. Data were collected on demographics, spinal cord imaging modalities, sacral ratio (SR) value, presence or absence of tethered cord, presence of associated vertebral anomalies, bowel and urinary control, surgical management for tethered cord, if present and outcome. Tethered cord was defined as termination of the conus medullaris below the L-2 endplate [3, 4]. Fecal and urinary control was assessed by the surgeon in children aged 3 years or more during the last clinic visit. Three years were used as the cutoff as this is the socially accepted age by which children are expected to be toilet trained. Fecal control was divided into three categories: full, voluntary bowel movements without soiling; partial, voluntary bowel movements with soiling; and no control (incontinence), absence of voluntary bowel movements. Urinary control was divided into three categories: full, complete diurnal and nocturnal control; partial, spontaneous voiding with dribbling between voiding episodes; and no control, requiring clean intermittent catheterization. The sacral ratio was calculated as described by Peña [19] and the results were divided into three groups: 0–0.4, 0.41–0.69 and $\geq$0.7 (normal). Findings were analyzed by descriptive statistics. This study was approved by the institutional review board.

Results

Types of ARMs and association with tethered cord

We identified 790 patients with ARMs, who underwent screening for TC during the study period. Tethered cord was identified radiologically in 285 (36%) patients. Of the reminder, findings were negative in 464 (58.7%) and equivocal in 41 (5.2%). Table 1 lists the types of ARM_S and the percentage of patients with tethered cord in each subgroup. The highest rates of tethered cord were found for vaginal fistula (100%), and cloacal extrophy (83.3%) and the lowest rates, for bulbar fistula (27%), perineal fistula (21.1%) and no fistula (16.6%). Generally, the more complex ARMs had higher rates of TC with two exceptions; vestibular fistula and rectal stenosis. Sixty-two patients had reoperations, primary malformation not specified, of whom 15 had a tethered cord.

Rectobladder neck fistula and tethered cord

By the time of the present study, a total of 52 patients with rectobladder neck fistula were screened for TC. Of these, 37 were followed by the senior author and formed the sample for further analysis. The median follow-up period of the cohort of 37 patients was 49 months (range 2–222 months). In 11 of 37 patients, the results of screening were positive for tethered cord. Ten patients were diagnosed by magnetic resonance imaging (MRI) and one by ultrasound. Of the 26 patients with negative findings, 16 were screened by MRI and 9 by ultrasound; in 1, the imaging modality was not specified. Thoracolumbar anomalies were found in 4 of the 37 patients with rectobladder neck fistula, all of whom were also positive for tethered cord (36.3% of the patients with tethered cord).

Relationship among tethered cord, sacral ratio, and outcome in patients with rectobladder neck fistula (Table 2)

The sacral ratio in the 37 patients with a rectobladder neck fistula who were screened for TC was $\geq$0.7 (normal) in 18, 0.41–0.69 in 12, and 0–0.4 in 7.

Table 1 Prevalence of tethered cord in patients with different types of ARMs

Type of ARM	Patients with TC, *n* (%)
Vaginal fistula	2/2 (100%)
Cloacal extrophy	20/24 (83.3%)
Rectal atresia/stenosis	7/10 (70%)
Complex malformations	23/39 (58.9%)
Vestibular fistula	54/117 (46.1%)
Rectobladder neck fistula	22/52 (42.3%)
Cloaca	72/199 (36.1%)
Prostatic fistula	28/102 (27.4%)
Bulbar fistula	20/74 (27%)
Perineal fistula	18/85 (21.1%)
No fistula	4/24 (16.6%)

Among the 18 patients with a sacral ratio of ≥ 0.7, 3 (16.6%) were found to have tethered cord. One was too young to be evaluated for fecal and urinary control status; one patient underwent tethered cord release (TCR) and is currently on clean intermittent catheterizations and bowel management for fecal incontinence, and one was followed expectantly, and is on bowel management, but data on urinary control data were unavailable.

Among the 12 patients with sacral ratio of 0.41–0.69, 4 (33.3%) were found to have a tethered cord. Three of them underwent TCR, none of whom has bowel control. Urinary control was achieved in two patients but data were missing for the third (a redo case). The fourth patient with a tethered cord did not undergo surgical release, and he has neither bowel nor urinary control. Table 2 shows the data for the other eight patients with normal cord.

Among the seven patients with SR 0–0.4, four (57.1%) had a tethered cord. Only one of them underwent TCR. He is fecally incontinent, on bowel management, but information on his urinary control was unavailable. The parents of another patient decided to leave the colostomy bag because the child has poor functional prognosis. Of the remaining two patients, one was too young for evaluation and one was on bowel management with no urinary control. Table 2 shows data of the other three patients with a normal cord.

Overall, the incidence of tethered cord rose with a decrease in sacral ratio. Patients with rectobladder neck fistula and tethered cord had dismal prognosis for voluntary bowel movements, regardless of the sacral ratio (Table 2). For patients with SR < 0.4, chances for voluntary bowel movement are nil and for urinary control very poor, whether they have TC cord or not.

All five patients who underwent TCR are fecal incontinent and on bowel management. Two patients required revision for retethering.

Discussion

Pediatric surgeons repeatedly raise the question of the optimal management of children with ARMs and tethered cord given that ARMs are often accompanied by preexisting impairment of bowel and bladder function.

Different studies of ARM_S have reported a wide range of overall rates of tethered cord of 9–52% [1, 14, 17, 18, 20]. The 36% rate found in the present study supports the practice of screening all patients with ARMs, regardless of type, during the newborn period. There is also conflicting evidence regarding the rate of TC with different types of ARM: some studies reported higher rates in association with the "lower" ARMs [1, 10, 20], whereas others found either equal rates in association with "lower" and "higher" ARMs [12],or an increase in the rate of tethered cord with an increase in ARM complexity [4, 15, 18, 21]. Our results show that incidence of TC varies according to the type of malformation and is increased the more complex the malformation, with exception of vestibular fistula (46%) and rectal stenosis (70%).

The incidence of TC in patients with vestibular fistula and absent vagina was found to be high (senior author practice), and higher incidence of TC was reported in 60% of patients with presacral mass and 57% of patients with sacral hemivertebrae [4].

In addition, in the subset of patients with rectobladder neck malformation, the incidence of tethered cord correlated

Table 2 Functional outcome in RBNF and TC (analysis adjusted to sacral ratio)

		SR Normal	SR 0.41-0.69	SR Poor
TC	Fecal control	3 0/8	4 0/8	4 0/8
	Urinary control	3 0/7	4 2/7	4 0/7
No TC	Fecal control	15 4/23 2/23 partial	8 1/23	3 0/23
	Urinary control	15 8/23	8 6/23	3 1/23

For the total 11 patients positive for tethered cord on screening (10 MRI, 1 US), 8 had available data on bowel control and 7 on bladder control

For the total 26 patients negative for tethered cord on screening (16 MRI, 9 US, 1 unspecified), 23 had available data on bowel control and urinary control

Numbers in square boxes indicate number of patients in the category of sacral ratio

with the sacral ratio: the lower the sacral ratio, the higher the rate of tethered cord.

Bowel control in patients with tethered cord

Several studies have evaluated bowel function in patients with ARM and tethered cord. Tsuda et al. identified nine patients with tethered cord out of 35 patients with ARMs. There were no differences between the patients with and without tethered cord on anorectal manometry and clinical evaluation of bowel function [15].

Other reported that among patients with ARMs, rates of fecal and urinary incontinence were higher in those with, than without tethered cord [4]. However, they noted that this finding may have been attributed to other factors such as the type of malformation and the status of the sacrum.

In one study, tethered cord was diagnosed in 10 of 110 patients with ARM, of whom only 1 was symptomatic. His symptoms were gait abnormalities and pain rather than poor bowel function [16].

To isolate the independent impact of tethered cord on bowel and bladder control, the analysis must adjust for the type of malformation and quality of sacrum, a key prognostic factor. However, the previous cited studies analyzed heterogeneous cohorts of patients with different types of malformations and different functional prognosis for bowel control and only a minority accounted for the sacral ratio. Therefore, in the present study, we concentrated on a specific type of malformation, rectobladder neck fistula, and analyzed outcome by specific subgroups of sacral ratio.

In our earlier study, we showed that rectobladder neck fistula carries a poor prognosis for bowel control. No bowel control was found in any of the patients with a low sacral ratio and in only 33.3% of those with a high sacral ratio [22]. Our current results show that patients with rectobladder neck fistula and tethered cord have dismal prognosis for voluntary bowel movement, unrelated to their sacral ratio status. For those patients with poor sacrum, the chances for urinary control were negligible, regardless of the presence or absence of tethered cord.

Impact of tethered cord release on bowel control

The lack of substantive data on the clinical course of ARMs patients with tethered cord makes it difficult to determinate the benefit of prophylactic untethering. Some investigators reported that release of a tethered cord in an asymptomatic patient may prevent the "expected clinical deterioration" and long-term neurological disabilities involving musculoskeletal and urological systems that are associated with the tethered cord [5]. This approach is supported by the variable onset of symptoms and gradually progressive course. Long et al. [14] studied 24 of 31 patients with ARMs and tethered cord who underwent surgical untethering. Those who were asymptomatic ($n=9$) remained so for an average follow-up time of 4.4 years. Among those who were symptomatic ($n=15$), motor deficits improved in only 60%, and bowel symptoms in 50%; there was no improvement in urological symptoms. The authors concluded that once symptoms of tethered cord become clinically apparent, they may be irreversible, particularly those associated with neurogenic bladder. Golonka et al. described 22 patients with ARM and TC all of whom underwent tethered cord release, 20 prophylactically and 2 after neurological symptoms developed. Two showed deterioration in neurological status and underwent repeated untethering within 2 years of their initial surgery. The authors pointed out that because ARMs often manifest with preexisting bowel and urinary deficits, it is difficult to distinguish the independent effect of each on bowel and bladder function, in patients with ARMs and tethered cord [10].

Morimoto et al. studied ten patients with ARMs and tethered cord with neurological deficits who underwent tethered cord release. None showed an additional neurologic deficit during 6 years of follow-up, supporting the use of an early, aggressive surgical approach in this patient group [11]. Muthukumar et al., in a study of seven patients with ARMs and tethered cord, noted an improvement in urinary incontinence following surgical intervention for the spinal anomaly, although fecal incontinence reported by one patient did not improve [12].

Others observed that an improvement in either bowel or urinary functions is not the rule following tethered cord release [1, 4]. Tuuha et al reported on 22 patients with ARM and TC. Seven of 22 patients underwent untethering; 3 prophylactic and 4 for neuro/motor function deficits. Following surgery, all four symptomatic patients had significant clinical improvement in their neuro/motor functions. However, bowel and urinary functions remained unchanged in all seven patients with a mean follow-up of 6.4 years. Fifteen patients with tethered cord remained asymptomatic with a mean follow-up of 2.7 years [1]. Accordingly, in none of 18 surgically treated patients with ARMs and tethered cord reported by Levitt et al. was there a significant improvement in bowel and urinary function following tethered cord release [4].

In a study of expectant conservative management, Kyrklund et al. reported no difference in long-term functional outcomes between patients with ARMs (specifically, rectovestibular and rectoperineal in females and rectourethral in males) and tethered cord and patients with the same type of ARM and a normal spinal cord [3]. In the study of Uchida et al., all 13 patients with ARMs and tethered cord, 10 symptomatic and 3 asymptomatic, showed improvement in orthopedic symptoms but not in bowel and urologic symptoms, following tethered cord release [20]. In Teo's study, all

17 patients with ARM and TC underwent surgical untethering, 10 asymptomatic and 7 symptomatic. While three of seven symptomatic patients had improvement in neurological deficit and post-operative urodynamic study, the other four showed no improvement. All ten asymptomatic patients remained asymptomatic [18].

Valentini et al., reported no improvement in sphincter function after surgery regardless of the type of malformation [23].

In the present study, none of the five patients who underwent untethering have bowel control and all require bowel management, regardless of their sacral ratio values.

Other factors also need to be taken into account by clinicians in the management of patients with ARMs and tethered cord. Although untethering is considered a straightforward procedure, it is not free of complications [24] such as cerebrospinal fluid leak (1.4–6%) [25–27] and wound infection (2.1–9.9%) [25, 28]. Additionally, there is a risk of retethering after the initial tethered cord release with the reported rates of 3.3–24% [25, 27, 29]. Yong et al. followed 152 patients of median age 4.5 years at the initial tethered cord release of whom 8.6% required repeated untethering within a mean time of 4.8 years [30]. Furthermore, neurologic deterioration following untethering was also reported [23, 31, 32].

Study limitations

This study is limited by the sample size which was too small for statistical analysis. The retrospective nature of the study led to incomplete data on some of the parameters studied. There may be a selection bias by a greater proportion of complex malformation being treated at a referral center. Also a selection bias by a greater proportion of patient with normal SR evaluated for TC, probably since patients with poor SR were screened less frequently in previous years. This study was focused on a subgroup of patients with rectobladder neck fistula. We plan to conduct similar studies on other types of malformations and tethered cord.

Conclusions

This study demonstrated that the rate of tethered cord in patients with ARMs increases with an increase in the complexity of the malformations, with the exception of rectovestibular and rectal stenosis. In patients with rectobladder neck fistula in particular, the rate of TC inversely correlates with sacral ratio value and is higher with lower sacral ratio. Patients with rectobladder neck fistula and tethered cord have dismal prognosis for voluntary bowel movement, unrelated to their sacral ratio status.

Many questions related to the management of all types of ARMs with asymptomatic tethered cord remain unresolved. Multicenter prospective outcome studies of tethered cord release and expectant management are needed to establish uniform guidelines. Pre- and post-operative urodynamic might be considered to detect neurological deficits.

Funding This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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