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Published in:
Journal of Pediatric Surgery

DOI:
10.1016/j.jpedsurg.2014.08.004

2015

Link to publication

Citation for published version (APA):

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Pelvic floor in females with anorectal malformations – findings on perineal ultrasonography and aspects of delivery mode

Pernilla Stenström a,b,⁎, Mette Hambraeus a,b, Einar Arnbjörnsson a,b, Ann-Kristin Örnö c

a Institution of Clinical Research, Lund University, Lund, Sweden
b Department of Pediatric Surgery, Skåne University Hospital, Lund, Sweden
c Department of Obstetrics and Gynecology, Skåne University Hospital, Lund, Sweden

A R T I C L E   I N F O

Article history:
Received 27 March 2014
Received in revised form 15 July 2014
Accepted 6 August 2014

Key words:
Anorectal malformations
Pelvic floor
Ultrasonography
Delivery mode

A B S T R A C T

Background: Advice on the mode of delivery to females born with anorectal malformation (ARM) is needed. The primary aim was to evaluate the anatomy of the pelvic floor muscles in females with ARM operated with posterior sagittal anorectal plasty (PSARP). The second aim was to correlate the extent of muscle defects to the bowel symptoms.

Methods: This interventional study with perineal 4D/3D ultrasonography describes the smooth muscles in the intestinal wall (neo-IAS), external sphincter, levators and anal canal using a muscle score (0–6 worst). The bowel symptoms were prospectively registered with Krickenbeck criteria score (0–7 worst).

Results: Forty females with different subtypes of ARM, median age 13 (4–21), were followed up regarding bowel symptoms. Seventeen were examined with ultrasonography. Bowel symptoms were similar for those examined with ultrasonography and those not, median score 5 and 3 (1–7) respectively, (p = 0.223, Fisher’s exact test). All the females had at least one muscular defect. There was no significant correlation between muscle defects and bowel symptoms (p = 0.094, Spearman’s correlation).

Conclusion: Females with ARM have considerable defects in the pelvic floor without any significant correlation to bowel symptoms. All women with ARM would benefit from individualized predelivery evaluations and caesarian section should be considered.

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The prevalence of anorectal malformations (ARM) among females is 15/100,000 live births. ARM includes a spectrum of anomalies in the pelvic floor with different subtypes of ARM [1]. All children born with ARM need a reconstructive operation in order to have the rectum located in the center of the sphincter complex. The reconstruction is mainly Posterior Sagittal Anorectal Plasty (PSARP), implemented worldwide in the late 80s [2].

The long-term outcome after PSARP is reported to depend especially on the ARM-subtype [3–5]. Among adolescent and adult females with different types of ARM, fecal incontinence is reported by 40–67% and lack of voluntary bowel control by 15–30% [3,6,7].

The length of follow up time of adult females with ARM operated on with PSARP is limited since the oldest PSARP-operated patients are 20–25 years old. Therefore few are of child bearing age and reports on deliveries are scarce. In view of this it is of interest to evaluate the anatomy of the pelvic floor among females with ARM.

Anal continence is dependent on the internal anal sphincter (IAS), the striated external anal sphincter (EAS) and M. levator ani [8,9]. In females with ARM the muscles and innervation deviate owing to the malformation. The original IAS is suggested to be rudimentary in the distal fistula [10,11] and can be resected during the PSARP [2,12]. Furthermore, during the PSARP, the EAS is divided both anteriorly and posteriorly, and M. levator ani is involved in the reconstruction [2,13].

Patients born with ARM in most cases lack the normal rectal properties such as a normal volume adaptation, the rectal sensibility might be compromised and a normal rectoanal inhibitory reflex is often missing [14–17]. Sacral and spinal anomalies may influence the bowel control [3,5,18]. Therefore bowel control might depend heavily on the muscle capacity.

When females with ARM become pregnant the question of mode of delivery may be an overlooked topic for some reason. Some females with ARM may have not been given enough information to be able to speak about their malformation or reconstruction [7]. The midwife or obstetrician may be unfamiliar with the diagnosis ARM [19]. Their life-long adaptation to the symptoms may lead to young females perceiving their fecal, urinary or gas incontinence as a “normal” condition and therefore this is not mentioned, unless specific questions are asked [7,20]. Furthermore, the scars after the PSARP reconstruction may not be obvious or thoroughly understood by those who are not familiar with ARM.

⁎ Corresponding author at: Department of Pediatric Surgery, Skåne University Hospital, 5–221 85 Lund, Sweden. Tel.: +46 46178373, +46730803600; fax: +46 46178120.
E-mail address: pernilla.stenstrom@med.lu.se (P. Stenström).

http://dx.doi.org/10.1016/j.jpedsurg.2014.08.004
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This study aims at contributing to the knowledge of the anatomy of the pelvic floor among young females with ARM operated on with PSARP. The primary aim of the study was to evaluate the pelvic floor muscles involved in the control of fecal continence in females with ARM operated on with PSARP using perineal ultrasonography. With this knowledge, better guidance with respect to mode of delivery could be given. The second aim was to correlate the severity of muscle anomalies to the severity of bowel symptoms, in order to evaluate if the patients’ history of bowel symptoms could provide information enough to support the decision of the mode of delivery.

1. Patients

The study included all females born with ARM between January 1990 until March 2009 who had been admitted to the Department of Pediatric surgery, a tertiary center which covers an area with 2 million inhabitants with free health care. ARM is divided into different subtypes according to the Krickenbeck classification which is based on the entry of the fistula from rectum [1]. Since we intended to describe the pelvic floor only after reconstruction with PSARP, limited PSARP or Posterior Sagittal AnoRectal Vaginal UréthроPlasty (PSARVUP) [13] those with other treatments, as anal stenosis and rectal atresia, were excluded. In the end 54 females born with the subtypes rectoperineal fistulas, rectovestibular fistulas and cloacas remained (Fig. 1a–c). Of these, 14 could not be included in the follow up because of death, severe syndromes or migration (Fig. 2). All of the remaining 40 females were followed up regularly at the department and 30 of these were invited to take part in the study which included perineal ultrasonography and registration of bowel symptoms. They belonged to the following groups:

1. Sixteen females, 14–21 years old, had their last medical consultation at the Department of Pediatric Surgery before transfer to adult medical care
2. Ten females, 10–13 years old, were planned for the regular pre-pubertal follow-up
3. Four females, 4–9 years old, were planned for general anesthesia for other reasons

The remaining 10 females were too young, 4–9 years old, and not planned for other general anesthesia for other reasons. Therefore they were not asked to participate in the examination with ultrasonography, but asked and agreed to being controls regarding bowel symptoms (Fig. 2).

2. Method

This is a prospective and descriptive interventional study. The study was conducted from July 2011 until May 2013. The patients were collected from the prospectively maintained database with all children with ARM in the region. The invitation to the study was made during the regular follow-ups at the Department of Pediatric Surgery.

2.1. Classification

The ARM were classified according to international standard of Krickenbeck classification [1] (Fig. 1a–c and Table 1).

2.2. Operation method

The reconstruction of anus was performed within the first months of life, and the standard procedure was PSARP [2]. During PSARP there is a incision performed in the midline of the pelvic floor, all the way from coccyx to vagina. The fistula and additionally 2 cm of rectum is resected. Rectum is mobilized to the center of the sphincter complex. The posterior rectum is fixed to the adapted posterior levators, the perineal body is built up and the external sphincter adapted both posteriorly and anteriorly. An anastomosis is established between full thickness rectum and the skin. Limited PSARP, used for perineal fistulas, is similar to PSARP but the incision in the midline is shorter and the operation more shalow. PSARVUP, used for cloacas, is a more complicated and extensive variant of PSARP, adding a separation and mobilization of urethra and vagina [2,13]. All the operations were performed or supervised by three pediatric surgeons with a colorectal pro. Diverting colostomy was used in the neonatal period for the females with cloaca and vestibular fistulas [21].

2.3. 4D/3D perineal ultrasonography

The ultrasonography examination of the pelvic floor was performed with the patient in the dorsal lithotomic position. The transducers (M6C, RSP6, system Voluson E8 GE) were held to allow a sagittal inspection of the levator ani and the anal sphincter complex [22,23]. All scans were saved in 3D. If the subjects were awake they were instructed to squeeze their pelvic muscles using a 4D/3D mode [22,24]. The scans were saved on a computer and analyzed off-line by a trained sonographer (AO).  

Fig. 1. Illustration of female types of anorectal malformation (ARM): Anatomical features of the three different subtypes of ARM in the present study: a) Rectoperineal fistula, b) Rectovestibular fistula, and c) Cloaca.
2.4. The description of muscles and anal canal

Descriptions focused on the deep and subcutaneous parts of the EAS, the neo-IAS, the levators and the distal part of the anal canal where the rectum is supposed to meet the skin. The evaluation of the EAS was made based on the possible diastase between the ends of the circular muscle anteriorly (Fig. 3b). Up to 15 degrees was considered to represent only scars after the PSARP, while a diastase >15 degrees was determined as a real defect in the muscle [15]. The neo-IAS i.e. the inner circular muscle layer, was measured and evaluated as fragmented or not. It was measured above the anastomosis or above the diastase between rectum and skin, described below (Fig. 3b).

The M. levator ani was described as with or without any visible disruptions. When the subjects were awake they were asked to squeeze and push.

The anal canal in some females with ARM clinically often diverts from others. A defect between rectum and the skin may be present, with a secondary “pocket” beneath the perineal skin. This discontinuation of both the intestinal wall and subcutaneous tissue was measured in mm. A distance of 5 mm was considered as scar, while >5 mm was considered as a defect (Fig. 3c).

In order to correlate the defects of the muscles to the severity of symptoms, a scoring of the muscles was performed with 1 point for every deviation (Table 3). Diastases in both the deep and superficial parts of EAS were considered as serious and thus given two points. The score ranged from 0 to 6 (6 = worst).

2.5. Bowel symptoms

The bowel symptoms were registered according to the Krickenbeck functional criteria. As suggested in the original article the outcome was measured before bowel management was introduced [1] (Table 2). The registration of bowel symptoms with the criteria is done routinely in the medical records and in the prospectively maintained database. The bowel symptoms were converted to a scoring system that had been used in a previous report [5]. The score ranged from 0-7 (7 = worst) (Table 2).

2.6. Investigation of sacrum and spinal cord

All the patients with ARM had been examined with spinal ultrasonography within 4 weeks after birth or with magnetic
resonance imaging (MRI) of the vertebra, sacrum and spinal cord. Patients with signs of sacral malformations or tethered cord on spinal ultrasonography were all further examined with MRI.

2.7. Anesthesia

All the females under 15 years of age were examined during general anesthesia performed with Propofol®, Sevoran gas® and Ultiva®. The older females were awake during the examination.

3. Statistical considerations

A statistician performed the statistical analyses. The comparisons of bowel symptoms were analyzed with Fisher’s two tailed exact test for dichotomous results and with the exact Kruskal-Wallis test for ordinal or numeric test variables. Spearman’s rank correlation was used for assessing the correlation between the non-parametric muscle score (0–6) and Krickenbeck symptom score (0–7) and between the defect distance in the anal canal and the severity of fecal incontinence.
Possible associations between fecal incontinence (Yes/No) and anatomical defects in neo-IAS or the whole EAS were analyzed by using Chi square tests. P-values below 0.05 were considered significant. Statistical computations were performed by using SPSS statistics software (PASW/SPSS software, version 18, IBM Corporation, Armonk, NY, USA).

3.1. Ethical consideration

The regional research ethics committee approved the study (registration number 2010/49). Every patient <18 years old had a written consent by their parents.

4. Results

4.1. Patients

All 40 females were followed up regarding their bowel symptoms. Out of the 30 who were asked to be examined with transperineal ultrasound, 17 (57%) agreed (Fig. 2). Five were awake during the examination.

The median age of all the females was 13.5 (4–21). The median age among those examined with ultrasonography and registration of bowel symptoms was 15 (4–21) and among those followed up without ultrasonography 11 (4–20). All the anorectal reconstructions were performed within the patients’ first 6 months of life, and the median time of follow-up postoperatively among all the females was 13 years (4–21). The frequency of the different subtypes of ARM among the females was similar to previously reported frequencies (Table 1).

4.2. Muscle components

The muscle defects in each patient and the total frequencies of the defects are presented in Table 4. None of the females had any rupture of the M. levator ani. All the females who were awake during the examination had a good control of the levators. The most frequent finding was fragmented IAS. In 11 females diastases in both the deep and superficial component of the EAS were identified (Figs. 3b and 4).

A majority of the patients had a gap of N 5 mm from the rectum to the skin anteriorly where also a lack of tissue under the superficial EAS and the skin was found (Fig. 3c).

4.3. Bowel symptoms

Fecal incontinence (Krickenbeck 1-3) was reported by 30/40 (75%) and constipation by 37/40 (93%) without bowel management. The use of regular bowel management with oral laxatives and/or enemas was reported by 31/40 (78%) with the aim to treat constipation and

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Table 4

<table>
<thead>
<tr>
<th>Krickenbeck follow-up</th>
<th>Females attending the study with perineal ultrasound</th>
<th>Females followed up without perineal ultrasound</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Total, n = 17</td>
<td>Total, n = 23</td>
</tr>
<tr>
<td></td>
<td>Perineal fistula Vestibular fistula Cloaca Perineal fistula Vestibular Fistula Cloaca</td>
<td></td>
</tr>
<tr>
<td></td>
<td>n = 9 n = 5 n = 3 n = 12 n = 10 n = 1</td>
<td></td>
</tr>
<tr>
<td>Voluntary bowel movements</td>
<td>Yes</td>
<td>10</td>
</tr>
<tr>
<td></td>
<td>No</td>
<td>7</td>
</tr>
<tr>
<td>Fecal incontinence</td>
<td>No</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td>Yes</td>
<td>15</td>
</tr>
<tr>
<td>Occasionally</td>
<td>Grade 1</td>
<td>3</td>
</tr>
<tr>
<td></td>
<td>Grade 2</td>
<td>10</td>
</tr>
<tr>
<td></td>
<td>Grade 3</td>
<td>2</td>
</tr>
<tr>
<td>Diet controlled</td>
<td>Grade 1</td>
<td>2</td>
</tr>
<tr>
<td></td>
<td>Grade 2</td>
<td>9</td>
</tr>
<tr>
<td>Laxative</td>
<td>Grade 3</td>
<td>6</td>
</tr>
<tr>
<td>Diet or laxative is not enough</td>
<td>Grade 1</td>
<td>5 (1–7)</td>
</tr>
<tr>
<td>Median Krickenbeck score 0–7 (7=worst)</td>
<td>5 (1–7)</td>
<td>5 (1–6)</td>
</tr>
</tbody>
</table>

The table illustrates those followed up with ultrasonography (US) and those without. n = number patients.

Total symptom score among those examined with US compared to without US (p = 0.223, Kruskal-Wallis).

Total symptom score compared between the different ARM-subtypes among those examined with US (p = 0.767), without US (p = 0.120), and for all patients (p = 0.485) (Kruskal Wallis).

Comparison of bowel symptoms between those examined with US and without US regarding:

- Voluntary Bowel Movements: (a) with US (p = 0.197) and (d) without US (p = 0.178) and for all patients (p = 0.071) (Pearson Chi-square).
- Fecal incontinence: (b) with US (p = 0.461), (e) without US (p = 0.276), and for all 40 patients (p = 0.378) (Kruskal-Wallis).
- Constipation (c) with US (p = 0.978), (f) without US (p = 0.085), and for all 40 patients (p = 0.343) (Kruskal-Wallis).

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Fig. 4. Perineal ultrasonography: Cross sectional image. Image of typical findings in females with anorectal malformation.
secondary over flow incontinence. For oral medication polyethylene glycol (makrogol) was used while sorbitol and/or saline were used for enemas.

The distribution of patients with voluntary bowel movements, fecal incontinence or constipation did not differ between the subtypes of ARM among those examined with or without ultrasonography. Neither were there any statistically significant differences found for the score between the groups (Table 5 and Fig. 5). Thus the group investigated with ultrasonography can be considered to be representative for all 40 females in terms of bowel function.

4.4. Sacral malformations and tethered cord

Sacral malformations were present in 12/40 (30%) and tethered cord in 5/40 (12.5%). The distribution of the sacral and spinal malformations was equal between those examined with perineal ultrasonography or not (Table 6). Among the 12 females with sacral malformations 9 reported any fecal incontinence (Krickenbeck grade 1–3) while 3 reported no fecal leakage. All 5 females with tethered cord had concomitant sacral malformation and they all reported daily fecal incontinence (Krickenbeck grade 2 or 3).

4.5. Correlations

The number of defects in the muscles and the severity of bowel symptoms did not correlate significantly even though the correlation coefficient was positive (Fig. 6). No significant correlation was shown between the diastase distance between rectum and skin and more severe (grade 1–3) fecal incontinence \( (p = 0.031, \text{Spearman's correlation}) \), fecal incontinence (Yes) and fragmentation of neo-IAS

![Fig. 5. Prevalence and comparison of bowel symptoms: Bowel symptoms (Yes/No) among females with anorectal malformation (ARM) followed up with perineal ultrasound and without, \( n \) = number of patients; \( ^* \) = Fisher's 2-sided exact test.](image)
5. Discussion

The whole EAS (p = 0.549, Chi-square test).

Females: attending the study with perineal ultrasonography

<table>
<thead>
<tr>
<th></th>
<th>Total n = 17</th>
<th>Perineal Fistula n = 9</th>
<th>Vestibular Fistula n = 5</th>
<th>Cloaca n = 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sacral malformation</td>
<td>Yes</td>
<td>6 (35%1)</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Tethered cord</td>
<td>Yes</td>
<td>2 (11%)</td>
<td>1</td>
<td>1</td>
</tr>
</tbody>
</table>

Females followed up without perineal ultrasonography

<table>
<thead>
<tr>
<th></th>
<th>Total n = 23</th>
<th>Perineal Fistula n = 12</th>
<th>Vestibular Fistula n = 10</th>
<th>Cloaca n = 1</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sacral malformation</td>
<td>Yes</td>
<td>6 (26%)</td>
<td>2</td>
<td>3</td>
</tr>
<tr>
<td>Tethered cord</td>
<td>Yes</td>
<td>3 (13%)</td>
<td>1</td>
<td>0</td>
</tr>
</tbody>
</table>

Numbers (n) and percentages (%).
1) and 2) p=0.786 and 0.650, respectively (Fisher’s exact test).

(p = 0.331, Chi-square test) respectively, to defects >15 degrees in the whole EAS (p = 0.549, Chi-square test).

5. Discussion

This is the first study to report the pelvic floor anatomy with focus only on females with ARM. It is also the first report using 4D/3D perineal ultrasonography as a method to describe the sphincter anatomy in females with ARM.

The main findings were that all females with ARM had at least one deviation from normal anatomy. There was no correlation between the number or types of defects and the severity of fecal incontinence. The diastase between rectum and skin anteriorly in the anal canal has never been illustrated before.

In 11/17 of the examined females the rectum did not reach the skin and a subsequent pocket under the skin and superficial EAS was found (Figs. 3c and 4b). One reason for this could be an insufficient mobilization of the rectum with a subsequent tension and diastase. Another reason could be a postoperative infection, with a secondary rupture or substantial defect. However, the clinical relevance of this finding remains unclear.

Fragmentation of neo-IAS was, surprisingly enough, noted in a majority of the females. We expected the neo-IAS to be unharmed, because of the mobilization of full thickness intestinal wall during the PSARP. The fragmentation may be owing to accidental damage during the PSARP or because of repeated dilatation of the neo-anus during the postoperative course. The relevance of the findings is unclear since the function of neo-IAS in ARM is unknown. Since there is some evidence that the original IAS may be located in the fistula, fistula saving surgery in ARM has been provoked [10,11]. In our cohort of patients the PSARP procedure was carried out according to the original PSARP and the latest recommendations, with resection of the fistula [2,12]. In the present study no correlation was found between fragmented neo-IAS and fecal incontinence. In contrast, previous studies on rectal sonography in children with ARM have shown a positive correlation between the number of scars in IAS and incontinence [14,15]. However these studies do not clarify whether the original IAS or neo-IAS has been measured. The same studies report EAS as incompletely adapted in a majority of the patients [14,15]. Those findings on EAS are similar to ours.

The levators in all females in this study were complete, even though some were separated. The role of the levators in healthy women is thought to contribute to fecal- and especially urinary continence [25]. Maybe the females with ARM and extensive scars in EAS, but acceptable continence, have managed to compensate the harmed neo-IAS and EAS with increased control of the intact levators. If so, the levators in women with ARM may play a very important role for continence.

The bowel symptoms among the females with ARM did not correlate to the status of the sphincter anatomy, even though there was a trend (Fig. 6). Only weak correlations between EAS disruptions and fecal incontinence have been shown previously [15,26]. We therefore conclude that only the medical history of a patient with ARM is not enough to decide the status of the sphincter complex. Secondary to this, the mode of delivery cannot be based on the patients’ history.

The outcome in ARM should be described with consideration to concomitant sacral and spinal malformations. The sacral ratio could unfortunately not be reported on since the older patients did not have any X-rays of sacrum performed. However, the status of sacrum and spinal cord was well controlled over the years with MRI. All the females with tethered cord reported daily fecal incontinence while the females with sacral defects reported diversity in severity of incontinence. The status of sacrum and spinal cord should be considered in all predelivery consultations in patients with ARM since it probably may influence the outcome also post delivery.

A possible bias in the study may be the selection of females who agreed to be examined with ultrasonography. However in the material analysis, there were no significant differences in bowel symptoms. The number of patients examined with ultrasonography is low and the three variants of ARM in the study may limit the general conclusions. The pelvic floor architecture may change over life time [27] and the heterogeneous age group in the present study, may be a limitation. Another factor to consider is that perineal ultrasound is new and not a technique usually reported, so comparisons with previous results from MRI and rectal ultrasound are difficult. Furthermore there are no standardized dimensions for the muscles in the anal channel or pelvic floor for females <18 years of age. Therefore the score of the muscles in this study was basic and broad.
Besides, the measurement of the sphincter was performed in 5 awake females and during anesthesia in 12 females. It is known that general anesthesia may influence the tonus of the sphincter muscles. However, the results for neo-IAS, EAS and the anal canal are not likely be influenced by the anesthesia. From this experience we think that 4D/3D perineal ultrasound in the future could be useful in clinical practice, also for children <12 years of age, without anesthesia, since it is non-invasive.

The physical outcome regarding bowel control and urinary leakage in gender mixed groups of patients with ARM and in males, has been reported to depend on concomitant spinal or sacral defects [3,5,18,28]. The focus in the present study was only the muscular status, but in a predelivery consultation it would be of importance also to consider possible sacral anomalies.

DURING VAGINAL DELIVERY IN PRIMIPARAS WITHOUT ARM 93% EXPERIENCE OBSTETRICAL TEARS AND 0.9–17.8% SUSTAIN A SPHINCTER RUPTURE [29–31]. If such ruptures were to occur during a vaginal delivery in a female with ARM, the reconstruction would be difficult since the anatomy already from before deviates from normal, as described in the present study. Additionally, 20–35% of the patients with ARM may have sacral and spinal cord deviations [3,5,32] which may make the patient with ARM even more vulnerable for secondary incontinence. After vaginal delivery 30% have avulsions in the levators and another 30% lack control of the levators [24,33]. Then, if a levator injury were to occur in a patient with ARM, whose fecal continence may depend on the levators, the risk of incontinence would probably be high. Further, in obstetric sphincter injuries the continence scores increase after a second vaginal delivery [34]. The usual recommendation to females with a previous sphincter rupture and anal incontinence is to deliver through caesarian section [35,36] although there are no good predictors on those with functional deficits and who will be worse after a vaginal delivery [37]. According to our results, the status of the EAS and neo-IAS in females with ARM is comparable to a previous sphincter rupture. Consequently vaginal delivery in females with all types of ARM, not only cloacas [19], is not to be recommended.

In conclusion the sphincter structures responsible for fecal continence in females with ARM divert considerably from a normal anatomy. The patients’ bowel symptoms do not indicate the status of the pelvic floor, and could not be the only information to rely on in the pre-delivery consultation. This study indicates that the pelvic floor in females with ARM seems to be damaged and therefore vulnerable to further damage that might be caused by vaginal delivery. Therefore all women with ARM would likely benefit from individualized predelivery evaluations and caesarian should be considered.

Acknowledgements

Gillian Sjödahl, Lexis English for Writers, Lund, Sweden, for linguistic revision of the manuscript and Håkan Lökvist biostatistician at the Competence centre for Clinical Research, Skåne University Hospital, LUND, Sweden, for statistical advice.

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