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Histology of the Terminal End of the Distal Rectal Pouch and Fistula Region in Anorectal Malformations

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OBJECTIVE: Until recently, surgeons have been posed with a dilemma—whether or not they should preserve the terminal end of the distal rectal pouch and the fistula region in anorectal malformations (ARMs). A detailed histological study of this region was conducted to establish a consensus for preserving or excising this region for reconstruction of ARMs.
METHODS: Histopathological examination using haematoxylin and eosin-stained sections of the terminal portion of the distal rectal pouch and proximal portion of the rectourogenital or rectoperineal connection was performed in 60 cases of high, intermediate and low ARMs.
RESULTS: Distorted internal sphincter was present in 93.3% of high, 90% of intermediate and 100% of low ARMs. The proximal fistula region was lined by transitional epithelium in 50% of cases, and anal glands were present in 83.3% and anal crypts in 68.3% of cases. The rectal pouch in the region of the internal sphincter and fistula was aganglionic in all cases.
CONCLUSION: This study shows that the terminal end of the distal rectal pouch and proximal fistula region possess distorted anal features with aganglionosis, and contradicts the recommendation that this region should be reconstructed in patients with malformations. [*Asian J Surg* 2008;31(4):211–5]

Key Words: anorectal malformations, distal pouch, fistula, histopathology

Introduction

The terminal end of the distal rectal pouch and fistula region have not been considered worthy of preservation until recently. Some investigators have now found manometric[1,2] and histological[3] evidence of features of a normal anus in this region, such as the presence of an internal sphincter, transitional epithelium, hypo- or aganglionosis, and anal glands and crypts. In the present study, we examined the histology of 60 cases to establish whether preservation or excision of the fistula region leads to better anal physiological function after reconstruction.

Methods

The tissue specimens consisted of 0.5–2.0 cm of the most distal part of the rectal pouch and the proximal portion of the rectourogenital or rectoperineal connection. The specimens were taken from patients undergoing a posterior-sagittal anorectoplasty (performed after a defunctioning colostomy or as a primary procedure without colostomy), an abdominoperineal pull-through, or anal transposition. A total of 60 patients were studied. Thirty had a high anorectal malformation (HARM), 16 men had intermediate ARM (IARM), four women had IARM

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(two with rectovaginal fistulae and two with rectovestibular fistulae), and 10 had anovestibular fistulae (AVF).

The distal end of the specimen was marked with black silk and it was immediately fixed in 10% formalin.[4] The specimen was then cut into transverse and longitudinal sections and processed further to prepare haematoxylin and eosin (H&E) stained paraffin sections, according to standard laboratory techniques for light microscopic studies. Detailed histological features around the proximal end of the fistula were evaluated (Tables 1 and 2) as follows:

- internal sphincter and its morphology;
- hypoganglionosis or aganglionosis;
- transitional epithelium;
- anal glands;
- anal crypts;
- subepithelial fibrosis;
- thickened nerve trunks;
- miscellaneous histopathological aberrations in the distal rectal pouch and fistula region.

Results and discussion

Until recently, most paediatric surgeons had assumed the absence of an internal sphincter in all HARMs and IARMs and in many low anal atresias. We found a distorted internal sphincter in 93.3% of cases (Table 2). However, normal morphology was seen in only 26.7%, and in 66.7% of cases, the internal sphincter was found to be disorganized or atrophic (Figures 1 and 2). In 6.7% of cases, no internal sphincter was visualized (Table 1).

Scott found a thickening of the circular muscle layer that resembles an internal sphincter in four females with low anal atresia.[5] Gans and Friedman[6] advocated preserving the rectal blind pouch, based on its histology. Yokoyama et al[7] showed distinct thickening of the circular and longitudinal muscle layers in the distal rectal pouch in two neonates with high anal atresia and rectourethral fistula. Meier Ruge and Holschneider[8] used the distal rectal pouch and fistulous region for ARM reconstruction.

Frenckner[1] has pointed out the significance of the rectourethral fistula as the location of the internal sphincter. Penninckx and Kerremans[2] were able to demonstrate an internal sphincter in both low and high anomalies, which was always localized in the region of the fistula. Lambrecht and Lierse[3] demonstrated the presence of a

Table 1. Features comparable to a normal anus around the fistula region: internal sphincter, transitional epithelium, anal glands, anal crypts, ganglion cells

Type of ARM	No. of cases	Internal sphincter		Transitional epithelium		Anal glands		Anal crypts		Ganglion cells	
		Present *n* (%)	Absent *n* (%)	Present *n* (%)	Absent *n* (%)	Present *n* (%)	Absent *n* (%)	Present *n* (%)	Absent *n* (%)	Present *n* (%)	Absent *n* (%)
HARM	30	28 (93.3)	2 (6.7)	14 (46.7)	16 (53.3)	23 (76.6)	7 (23.4)	20 (66.6)	10 (33.3)	–	30 (100)
IARM (males)	16	14 (87.5)	2 (12.5)	10 (62.5)	6 (37.5)	13 (81.3)	3 (18.8)	12 (75.0)	4 (25.0)	–	16 (100)
IARM (females)											
RVg fistula	2	2 (100)	–	–	2 (100)	2 (100)	–	1 (50.0)	1 (50.0)	–	2 (100)
RVs fistula	2	2 (100)	–	–	2 (100)	2 (100)	–	1 (50.0)	1 (50.0)	–	2 (100)
AVF	10	10 (100)	–	6 (60.0)	4 (40.0)	10 (100)	–	7 (70.0)	3 (30.0)	–	10 (100)
Total	60	56 (93.3)	4 (6.7)	30 (50.0)	30 (50.0)	50 (83.3)	10 (16.7)	41 (68.3)	19 (31.7)	–	60 (100)

ARM = anorectal malformation; HARM = high ARM; IARM = intermediate ARM; RVg = rectovaginal; RVs = rectovestibular; AVF = anovestibular fistula.

Table 2. Morphology of the internal sphincter

Type of ARM	No. of cases	Morphology of internal sphincter		
		Normal *n* (%)	Disorganized or atrophic *n* (%)	Not visualized *n* (%)
HARM	30	10 (33.33)	18 (60.0)	2 (6.7)
IARM (males)	16	6 (37.5)	8 (50.0)	2 (12.5)
IARM (females)				
RVg fistula	2	–	2 (100.0)	–
RVs fistula	2	–	2 (100.0)	–
AVF	10	–	10 (100.0)	–
Total	60	16 (26.7)	40 (66.7)	4 (6.7)

ARM = anorectal malformation; HARM = high ARM; IARM = intermediate ARM; RVg = rectovaginal; RVs = rectovestibular; AVF = anovestibular fistula.

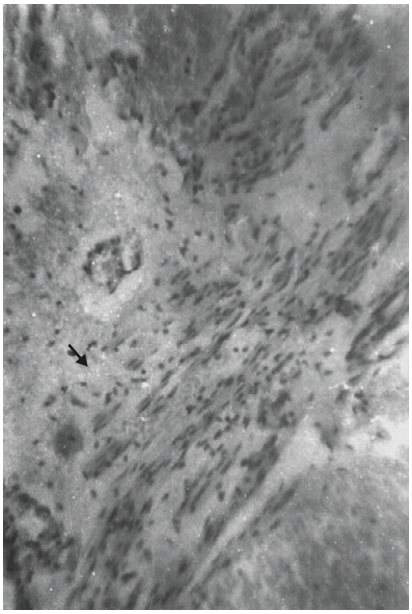

Figure 1. Disorganized and fragmented smooth muscle cells (internal sphincter) with capillaries and areas of haemorrhage (haematoxylin & eosin, 150×).

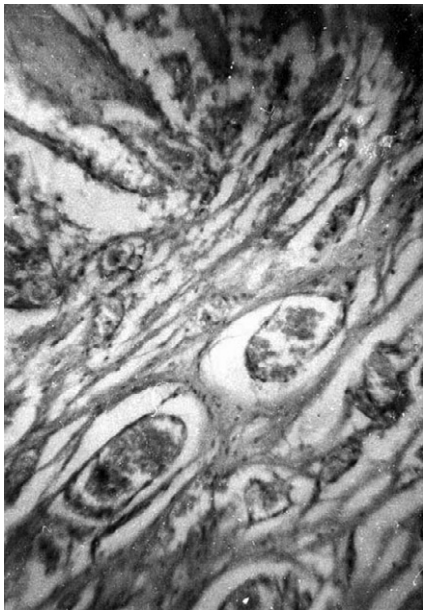

Figure 2. Dilated blood vessels in the rectal muscle layer associated with atrophy of muscles (haematoxylin & eosin, 150×).

normal internal sphincter in all ARMs. They showed that the internal sphincter was always localized around the internal fistula orifice in piglets; and in those animals without a fistula, the internal sphincter was at the deepest point of the rectal pouch. If the fistula originated from the deepest point of the rectal pouch, the internal sphincter was also localized at the deepest point. However, the fistula frequently originated further cranially from the deepest point of the rectal pouch and in such a situation, it was often not in the midline. In these cases, the internal sphincter was located eccentrically in the rectal pouch around the fistula region.[3]

The internal sphincter is the most important factor in the anorectal resistance barrier and is thus essential for continence. The term fistula seems incorrect and the bowel opening should rather be called an ectopic anus. This was suggested 30 years ago by Gans and Friedman[6] and Bill and Johnson.[9] After embryological studies, they presented a theory of incomplete migration of the distal bowel segment that results in the rectal opening not reaching its correct position in the perineum. They also found histological similarities between the fistulous connections in ARMs and the normal anal canal and proposed that no part of the terminal bowel should be

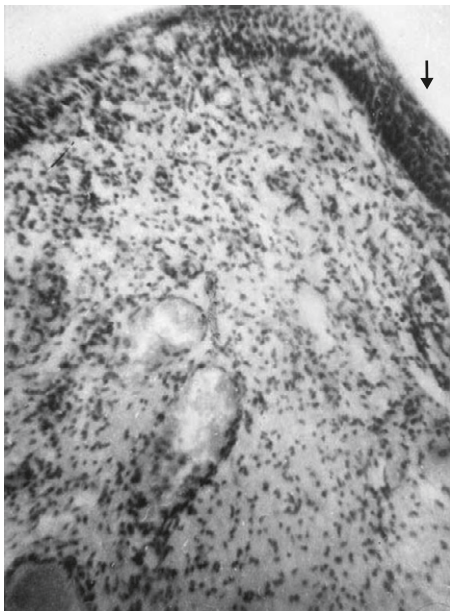

Figure 3. Squamous epithelium (arrow) with subepithelial fibroblastic proliferation, inflammatory cells and congested vessels (haematoxylin & eosin, 150×).

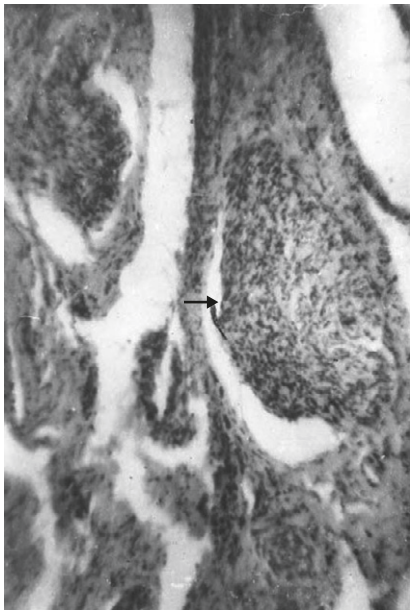

Figure 4. Hypertrophic nerve bundles (arrow) (haematoxylin & eosin, 150×).

Table 3. Histological changes in the fistula region: subepithelial fibrosis, thickened nerve trunks

Type of ARM	No. of cases	Subepithelial fibrosis		Thickened nerve trunks	
		Present, *n* (%)	Absent, *n* (%)	Present, *n* (%)	Absent, *n* (%)
HARM	30	30 (100.0)	–	12 (40.0)	18 (60.0)
IARM (males)	16	1 (75.0)	4 (25.0)	12 (75.0)	4 (25.0)
IARM (females)					
RVg fistula	2	2 (100.0)	–	2 (100.0)	–
RVs fistula	2	2 (100.0)	–	2 (100.0)	–
AVF	10	8 (80.0)	2 (20.0)	8 (80.0)	2 (20.0)
Total	60	54 (90.0)	6 (10.0)	36 (60.0)	24 (40.0)

ARM = anorectal malformation; HARM = high ARM; IARM = intermediate ARM; RVg = rectovaginal; RVs = rectovestibular; AVF = anovestibular fistula.

resected unnecessarily, which has been contradicted subsequently by Meier Ruge and Holschneider.[8]

The presence of a rectoanal inhibitory reflex, which indicates a functional internal sphincter in the fistula region, has been shown by pre-, peri- and postoperative manometry.[2,10] In addition to the internal sphincter, other features of a normal anus in the fistula region include absence of ganglion cells and the presence of transitional epithelium and glands and crypts. We did not find transitional epithelium in any of our cases (Table 2). The transitional epithelium of the anal region (fistula area) contains sensorial endings useful in the control of motor function and, hence, continence is not expected as normal if it is preserved. Crypts were found in 68.3% of our cases. These are the sites of opening for anal glands, which were present in 83.3% of our cases (Table 2), but they have no functional value for anal continence.

The marked subepithelial fibrosis in the transitional epithelial zone is probably secondary to faecal stasis, and in men, to urine reflux from the urethra to the blind pouch.[10] We found subepithelial fibrosis (Figure 3) in 90% of our cases (Table 3).

Thickened nerve trunks (Figure 4) in the distal rectal pouch were found in 60% of our cases, most commonly in IARMs (Table 3). Meier Ruge and Holschneider[8] observed oligoneural hypoganglionosis of the myenteric plexus proximal to the anal floor, also most commonly in IARMs. Both of these findings are consistent with a Hirschsprung-like picture in the distal rectal pouch, which is therefore strongly recommended to be excised for reconstruction.

Table 4. Miscellaneous histopathological findings in the distal rectal pouch and fistula region

Histological findings	HARM ($n=30$)	IARM (males) ($n=16$)	IARM (females)		AVF ($n=10$)	Total ($n=60$)	%
			RVgF ($n=2$)	RVsF ($n=2$)			
Increased blood vessels	8	2	–	–	2	12	20.0
Submucosal haemorrhage	4	2	–	–	2	8	13.3
Thrombosis of blood vessels	2	–	–	–	–	2	3.3
Submucosal lymphoid hyperplasia	8	–	–	–	2	10	16.7
Chronic inflammatory cells	2	–	–	–	2	4	6.7
Mucosal ulceration	2	–	–	–	2	4	6.7
Submucosal oedema	–	4	2	–	–	6	10.0
Disrupted muscularis mucosa	4	2	–	–	–	6	10.0
Thickened muscularis mucosa	2	–	–	–	–	2	3.3
Striated muscles	2	2	–	–	2	6	10.0

ARM = anorectal malformation; HARM = high ARM; IARM = intermediate ARM; RVgF = rectovaginal fistula; RVsF = rectovestibular fistula; AVF = anovestibular fistula.

The occurrence of constipation in patients whose fistula region is preserved is clearly related to the presence of a distorted internal sphincter, subepithelial fibrosis and aganglionosis. This leads to increased anal resting pressure generated by preservation of this fistula region, together with decreased rectal sensitivity (due to partial sensory denervation of the distal rectum). Congenital rectal dilatation may predispose patients to the development of constipation and anal incontinence.[11]

We observed several other histological aberrations in the distal rectal pouch (Table 4; Figure 2). It is likely all these point to a state of chronic inflammation and injury in the rectal blind pouch, which may be caused by faecal impaction and stasis within the distal pouch and/or reflux of urine into the pouch.

We also observed the presence of disrupted muscularis mucosa (10%), thickened muscularis mucosa (3.3%) and striated muscles (10%) in the distal rectal pouches (Table 4). These are indicative of the histological aberrations present embryologically, and may be implicated in the pathogenesis of postoperative constipation, if this part of the rectal pouch is preserved.

Hence, the fistula region is probably responsible for constipation because of abnormal morphology and innervation. The higher incidence of chronic constipation after surgical correction of all variants of ARM is possibly the consequence of these structural aberrations. This observation supports the concept of excising the distal rectal pouch and fistula region to achieve better continence.

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