Original Article

Histology of the Terminal End of the Distal Rectal Pouch and Fistula Region in Varying the Severity of Anorectal Malformations: Is it Useful?

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Abstract

Aims: To Study the histology of the terminal end of distal rectal pouch and fistula region in cases of anorectal malformations (ARMs) and to get an insight on the usefulness of excising or preserving this region during its reconstruction. **Materials and Methods:** This was a prospective observational study of 20 consecutive cases of ARMs that underwent posterior sagittal anorectoplasty (PSARP) in our hospital over 6 months' period. The histopathological evaluation of the terminal end of the distal rectal pouch and fistula region in all cases with ARM was done. Complicated and redo cases were excluded from this study. Tissue specimen of about 0.5–1.0 cm from the most distal part of the rectal pouch and close to the fistula region was taken. Biopsy specimens were obtained from all patients undergoing a PSARP performed after a defunctioning colostomy or as a primary procedure without colostomy, and histopathological evaluation was done in all cases. Further, the internal sphincter and its morphology, hypoganglionosis or aganglionosis, anal glands and crypts, thickened nerve trunks, and other miscellaneous histopathological aberrations were studied. Based on these histological findings, conclusions were derived whether to preserve or excise this region during ARM reconstruction. **Results:** Out of the 20 ARM patients included, 12 patients (60%) were male and 8 (40%) were female. An internal sphincter was identified in all the patients. However, the smooth muscle bundles were disorganized in all the 20 patients (100%). While ganglion cells were absent in 90% cases, hypertrophic nerve bundles were a common histological finding (90% of patients). The abnormal mucosal finding was also noted in majority of the patients (75%). **Conclusions:** An atrophic or disorganized internal sphincter, absent ganglion cells, and abnormal anal mucosal findings in majority of these patients on histology would justify its excision during PSARP. However, further follow-up of these ARM patients in whom the terminal end of the distal rectal pouch and the fistula region is excised or retained is recommended.

Keywords: Anorectal malformations, histology, rectum

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Introduction

It is a matter of debate among surgeons worldwide whether or not they should preserve the terminal end of the distal rectal pouch and the fistula region in anorectal malformations (ARMs). Some investigators have found manometric and histological evidence of the feature of the normal anus in this region, such as the presence of internal sphincter, transitional epithelium, hypoganglionosis or aganglionosis, and anal glands and crypts. Few surgeons have used this distal rectal pouch and fistula region for ARM reconstruction.[1-9] While others have reported distorted histological features in this region and contradict the recommendations that this region should be reconstructed in patients with ARM.[10] The present study is aimed to justify whether preserving or excising this region would lead to better functional outcomes after the reconstruction of varying severity of ARMs based on the histological evidence of tissue

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derived during its reconstruction. The presence of normal anal mucosa along with the normal internal sphincter in this region would rationalize its preservation during posterior sagittal anorectoplasty (PSARP) while an atrophic or disorganized or absent internal sphincter and abnormal anal mucosal findings would justify its excision during its reconstruction.

Materials and Methods

A prospective observational study of 20 consecutive cases of ARMs, including both male and female patients, operated over 6 months' study period. The histopathological examination of the terminal end of the distal rectal pouch and fistula region in all cases was done. Complicated and redo cases were excluded from this study. A detailed history of each patient was taken with the special emphasis on the type of ARM and whether associated with other congenital malformations such as cardiac, urogenital, gastrointestinal, vertebral, or VACTERL (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities). Thorough perineal examination including the number of openings, anal dimple, gluteal cleft, buttocks, genitalia and routine investigations, including hemogram, kidney function test, and urinalysis, including microscopy were done in all the patients. Other radiological studies, including X-ray abdomen, ultrasonography of the kidney, ureter, and bladder, echocardiography, and distal colagram study [Figure 1] were also done in all patients with high ARMs. The tissue specimen [Figure 2] of about 0.5–1.0 cm from the most distal part of the rectal pouch and close to the fistula region was taken. The biopsy specimens were obtained from all patients undergoing a PSARP that is performed after a defunctioning colostomy or as a primary procedure without colostomy. Histopathological examination using hematoxylin and eosin (H and E)-stained sections were performed in all cases. A detailed histological study was conducted on the formalin-fixed specimen at the proximal end of the labeled fistula region under the light microscope after staining with H and E.[11] Furthermore, internal sphincter and its morphology, hypoganglionosis or aganglionosis, thickened nerve trunks, mucosal abnormalities, and other miscellaneous histopathological aberrations were studied. Based on these histological findings, conclusions were derived whether to preserve or excise this region during ARM reconstruction. All cases were followed after surgery, and detailed history was taken regarding bowel habits, including a history of constipation, anal incontinence, or fecal soiling.[12,13] Data were simultaneously entered into a detailed pro forma during the study period and further evaluated. The Institutional Ethical Clearance was obtained for the study.

Results

Out of the 20 ARMs patients included, 12 patients were male (60%) and 8 (40%) were female. All male patients had high ARMs, whereas females had low ARMs. An internal sphincter was identified in all the patients [Table 1]. However, the smooth muscle bundles were disorganized in all the patients [Figure 3].

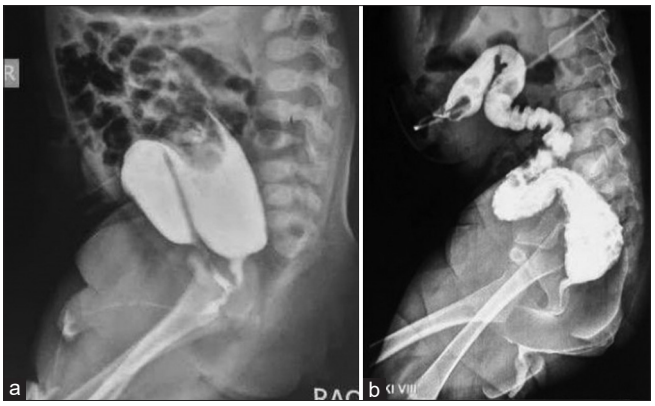

Figure 1: (a and b) Distal colagram showing contrast in the fistula region and distal rectal pouch in male (a) and female (b) patients

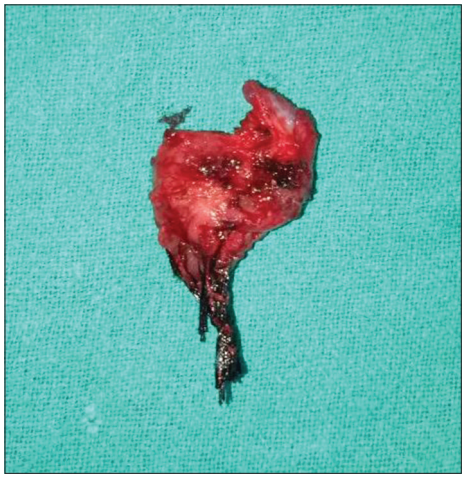

Figure 2: Specimen of the fistula region sent for histopathology in a female patient (distal end of the fistula marked with black silk threads)

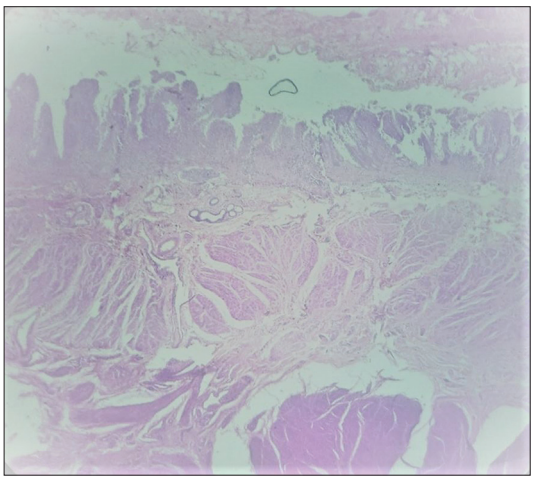

Figure 3: Histological findings (hematoxylin and eosin stain): Muscularis propria showing disarray of muscle bundles of inner circular and outer longitudinal layers

Table 1: Histopathological findings in terminal end of the distal rectal pouch and fistula region in our study patients

Gender	Type of ARM	Mucosal findings	Muscle morphology (IS)	Presence of ganglion cells	Hypertrophic nerve bundles	Miscellaneous histological findings
Female	Low	Normal	Disorganized	Absent	Present	-
Male	High	Focal ulcerations	Disorganized	Absent	Present	-
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Male	High	Focal ulcerations	Disorganized	Present (occasional)	Present	Submucosal chronic inflammation
Female	Low	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Male	High	Focal ulcerations	Disorganized	Present++	Present	-
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Female	Low	Normal	Disorganized	Absent	Present	-
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Male	High	Focal ulcerations	Disorganized	Present++	Present	Submucosal chronic inflammation
Female	Low	Focal ulcerations	Disorganized	Present (occasional)	Present	-
Male	High	Focal ulcerations	Disorganized	Absent	Absent––	Submucosal chronic inflammation
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Female	Low	Normal	Disorganized	Absent	Present	-
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Female	Low	Focal ulcerations	Disorganized	Absent	Absent––	-
Female	Low	Normal	Disorganized	Absent	Present	Submucosal chronic inflammation
Male	High	Focal ulcerations	Disorganized	Absent	Present	Submucosal chronic inflammation
Female	Low	Normal	Disorganized	Absent	Present	-

ARM: Anorectal malformation, IS: Internal sphincter

Abnormal mucosal and submucosal findings, namely focal mucosal ulcerations, submucosal edema, and chronic inflammation were seen in majority (>50%) of the patients [Figure 4].

Ganglion cells in the myenteric plexus were absent in 16 patients (80%) [Figure 5]. Occasional ganglion cells were seen in two patients. In the remaining two patients (both male ARMs), the ganglion cell was identified. Hypertrophic nerve bundles were a common histological finding associated with 90% of patients [Figure 6].

Discussion

Frenckner has documented the importance of the rectourethral fistula as the location of the internal sphincter in 1985.[1] Penninckx and Kerremans in 1986 revealed that the internal sphincter in both low and high anomalies was always localized in the region of the fistula.[2] Lambrecht and Lierse also confirmed the presence of a normal internal sphincter in all ARMs which 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.[3] If the fistula originated from the deepest point of the rectal pouch, the internal sphincter was also localized at the deepest point. 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. However, 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 the continence. The term fistula seems incorrect and the bowel opening should rather be called an ectopic anus as was suggested by Gans and Friedman as well as Bill and Johnson.[5,7] They presented a theory of incomplete migration of the distal bowel segment that resulted 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 resected unnecessarily that was contradicted subsequently by Meier-Ruge and Holschneider.[9]

Most pediatric surgeons had affirmed the absence of an internal sphincter in ARMs. Furthermore, Scott found a thickening of the circular muscle layer resembling an internal sphincter in four females with low anal atresia.[4] In addition, Gans and Friedman further advocated the preservation of the rectal blind pouch based on the histological finding.[5] Yokoyama *et al*. demonstrated distinct thickening of the circular and longitudinal muscle layers in the distal rectal pouch in two neonates with high anal atresia and rectourethral fistula.[6] Meier-Ruge and Holschneider used the distal rectal pouch and fistulous region for ARM reconstruction.[9]

The presence of a rectoanal inhibitory reflex, which indicates a functional internal sphincter in the fistula region, has been shown by preoperative, peri-operative, and postoperative manometry.[2,11] In addition to the internal sphincter, other features of a normal anus in the fistula region include the absence of ganglion cells and the presence of transitional epithelium and glands and crypts.

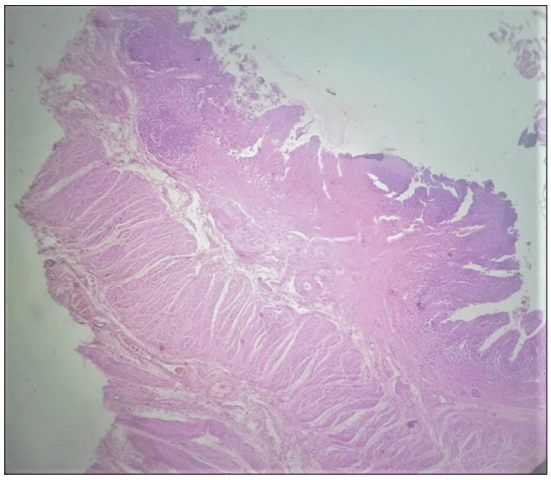

Figure 4: Histological findings (hematoxylin and eosin stain): Focal mucosal ulcerations

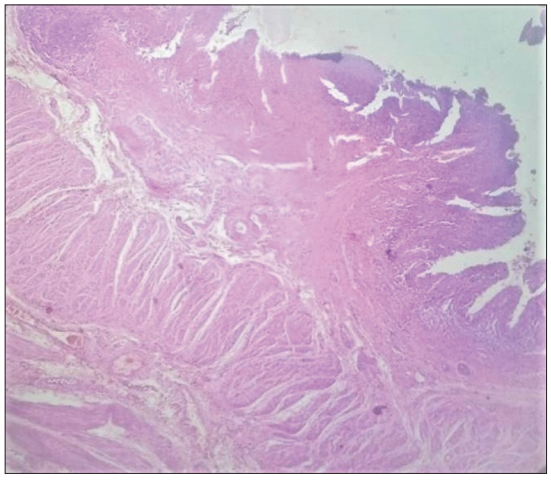

Figure 5: Histological findings (hematoxylin and eosin stain): Absence of ganglion cells in myenteric plexus in a distal rectal pouch and fistula region

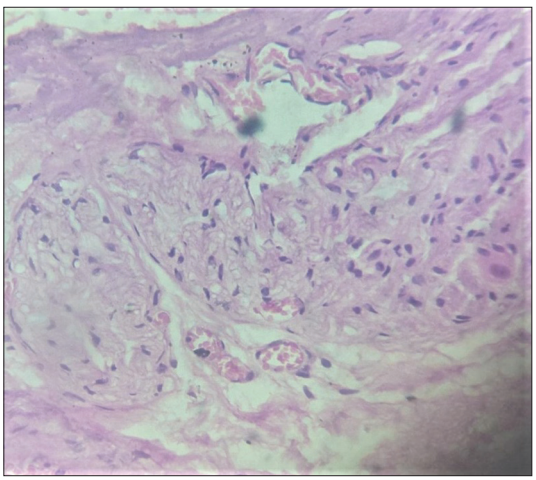

Figure 6: Histological findings (hematoxylin and eosin stain): Presence of hypertrophic nerve bundles in the distal rectal pouch

Thickened nerve trunks [Figure 4] in the distal rectal pouch were found in 90% of our cases. Meier-Ruge and Holschneider observed oligoneural hypoganglionosis of the myenteric plexus proximal to the anal floor.[9] 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 the reconstruction. 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 the 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.[12,13]

Other histological aberrations in the distal rectal pouch are due to a state of chronic inflammation and injury in the rectal blind pouch, which may be caused by fecal impaction and stasis within the distal pouch and/or reflux of urine into the pouch. We also observed the presence of focal mucosal ulceration in majority of our patients [Figure 2]. These histological aberrations 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. These structural aberrations are histologically confirmed by absent ganglion cells, hypertrophic nerve bundles, mucosal and submucosal alterations, namely ulcerations, edema, chronic inflammation, and subepithelial fibrosis.

Conclusions

An atrophic or disorganized internal sphincter, absent ganglion cells, and abnormal anal mucosal findings in the majority of these patients on histology would justify its excision during PSARP. However, further follow up of these ARM patients is needed in whom the terminal end of distal rectal pouch and the fistula region is excised or retained.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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