

Tailgut Cyst in an Infant with Imperforate Anus: a Case Report

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Abstract

Background: Tailgut cyst (TGC) is a rare congenital lesion that originates from remnants of the embryonic post-anal gut. It presents as a multilocular presacral mass mainly in young women. Microscopically, the cyst lining is composed of different types of epithelium such as stratified squamous, transitional, or glandular.

Case Presentation: We present a term female newborn referred to our hospital for evaluation and management of imperforate anus. During dissection of the presacral space to release the rectum, a multicystic mass adherent to the distal part of rectum was detected and completely excised. Histopathology confirmed the TGC diagnosis.

Conclusion: TGC is a very rare lesion, but it should be considered in differential diagnosis of any presacral mass, even in infancy. Complete excision is the preferred treatment and can be done more easily neonatally or in infancy.

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Introduction

Tailgut cyst (TGC), first described by Middeldorpf in 1885, as a rare congenital, embryonic hindgut remnant lesion^[1], occurs extremely rarely in infancy. It usually presents as a multiloculated cyst in the retrorectal space in adults, mostly young women^[2]. Its histologic diversity can confuse the pathologists seeking a definitive diagnosis^[1]. Herein we report a rare case of TGC in an infant with imperforate anus.

Case Presentation

A 3400-g female term newborn was born by

normal vaginal delivery and had Apgar scores 9/10 at 1 and 5 minutes. There was no complications during pregnancy and delivery. She was referred to our hospital on the first day of life for evaluation and management of imperforate anus. Perianal physical examination showed imperforate anus without any fistula and normal external female genitalia. Physical as well as neurological examination was normal. On the second day of life she underwent proximal sigmoid colostomy with biopsy for evaluation of ganglion cells that was normal in pathology. At 10 weeks of age, she underwent a distal colostogram to look for fistulas and then underwent a posterior sagittal anorectoplasty. During presacral exploration for distal part of the rectum, a well-demarcated cystic mass adherent to the distal part

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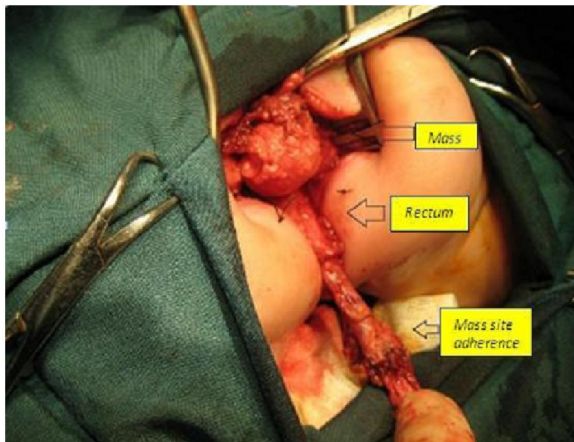


Fig. 1: Well-demarcated cystic mass in presacral space, adherent to distal part of the rectum

of rectum posteriorly measuring 4×3cm was detected (Fig. 1). The mass was completely excised with distal rectum and PSARP was done.

Pathologic examination of resected mass revealed a multilocular cyst lined by squamous, transitional and pseudostratified epithelium with goblet cells (Fig. 2). The underlying stroma was composed of fibrous tissue containing scattered bundles of smooth muscle fibers with infiltration of foamy histiocytes. These findings were consistent with TGC.

The infant recovered without complication and was discharged on the sixth postoperative day. The patient had an uneventful postoperative course and to date, 14 months after surgery, she is well, with no evidence of disease or any complications.

Discussion

TGCs (retrorectal hamartoma, cyst of postnatal intestine, rectal cyst, myoepithelial hamartoma of the rectum, tailgut vestiges) are rare congenital developmental lesions that are thought to be derived from remnants of the embryonic post-anal gut^[2]. During days 28-35 of embryonic development, the embryo possesses a true tail, which is caudal to the site of subsequent formation of the anus. It constitutes the tailgut or postnatal gut, which regresses by the 8th week of gestation. It is hypothesized that failure to regress of the embryological tailgut results in formation of TGC^[4]. They occur mostly in the retro-rectal/presacral space but have also been reported in the perianal, anorectal or posterior sacral regions^[5].

They occur most frequently in young adult women (3:1 female to male ratio)^[2]. Although they have been reported at any age, their diagnosis in a neonate like ours is extremely rare. Hjermstad and Helwig in their series of 53 cases of TGCs collected over a 35-year period, reported only one case in a 4-day infant^[2]. We know only of two other cases of neonatal TGC. One reported by Oh et al presented as a huge sacrococcygeal mass while another case had anal stenosis^[4,6]. We know of no TGC case associated with imperforate anus.

There is an association between TGCs and sacrococcygeal abnormalities. Hjermstad and Helwig suggest that a stimulus preventing the complete regression of the tailgut may also block closure of vertebral axis in this region and cause

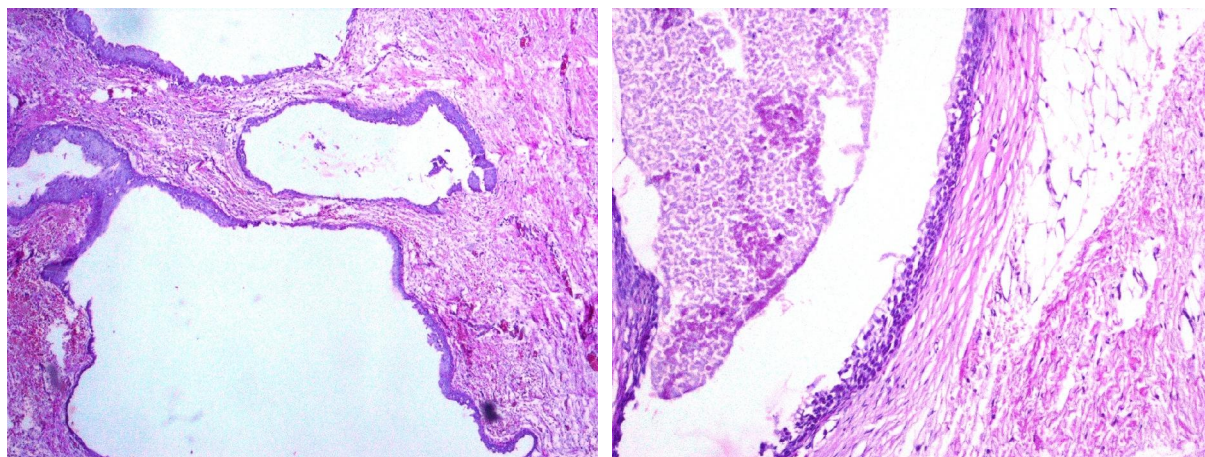


Fig. 2: A. Multilocular cyst lined by squamous, transitional and columnar epithelia (left). B. Pseudostratified columnar epithelium with goblet cells and underlying fibrous stroma (right).

sacral anomalies^[2]. This theory may also explain occurrence of imperforate anus in our case.

Although TGCs are congenital lesions, they are usually not diagnosed until adulthood^[2]. They are often asymptomatic and discovered incidentally (such as ours during anorectoplasty) but can cause symptoms related to local mass effect such as discomfort on sitting, painful bowel movements or urinary obstruction^[2-4]. Infections with fistulization and bleeding are the major complications reported specifically in adults^[2].

Due to their location, TGCs are palpable on rectal examination^[7]. Transrectal ultrasonography, computed tomography (CT) and MRI are preoperative diagnostic tools that can show the exact location and its cystic nature^[8]. However other developmental cysts have imaging findings similar to TGC, and the definite diagnosis relies on histological examination.

Grossly, this lesion can be unilocular or multilocular (50%), ranging in size from 1cm to 15cm and its content is variable from clear fluid to dense mucus. Histologically, the lining epithelium of cysts contain transitional and/or glandular-type (columnar) epithelium, with or without stratified squamous components and an underlying stroma with fibrous connective tissue and scattered bundles of smooth muscle fibers. An important distinctive feature is absence of well-defined muscular layer with nerve plexus and serosa^[2,3].

Regarding the complex embryology of presacral space there is a wide range of differential diagnoses including teratoma, epidermoid and dermoid cyst, rectal duplication cyst, neurenteric cyst, anal duct or gland cyst and anterior meningocele^[2,3]. Sacrococcygeal teratoma which is the most common neoplasm of the newborn, consists of mixed cystic and solid components and also contains skin adnexae, neural elements, and heterologous mesenchymal tissues that are never seen in TGC^[9]. Epidermoid cysts are lined only by stratified squamous epithelium and lack smooth muscle fibers in the wall. In addition, dermoid cysts contain skin appendages (e.g., hair follicles and sweat glands). Rectal duplication cysts are usually unilocular with prerectal location, lined by an intestinal or respiratory epithelium and have well-defined muscle layers with myenteric plexus. Anal duct or gland cysts can be distinguished by their lower location and their close proximity to anal sphincter and not being in the

retrorectal/presacral space. Neurenteric cysts differ from tailgut cysts in their histopathologic analysis in that they contain a well defined lamina propria and a more mature mucosa of endodermal origin (eg, intestine, bladder). Anterior sacral meningocele is associated with a sacral defect^[3,9,10].

Malignant change is a rare event in TGCs, but adenocarcinomas, carcinoid tumors, neuroendocrine carcinomas, endometrioid carcinoma, adenosquamous carcinoma, squamous cell carcinoma and sarcoma have been reported. The majority are adenocarcinomas and carcinoid tumors^[11-14]. Moreira et al showed that malignant transformation in the tailgut cyst is associated with mutation in the tumor inhibitory gene *p53* and suggested that dysplasia-carcinoma sequence in a tailgut cyst is similar to the one described in colonic carcinomas based on the analysis of two cases^[1].

Because of its potential for infection, occurrence of recurrent perianal fistulas, and the possibility of malignant transformation, complete surgical resection for all TGCs is recommended^[3,15]. It seems that the excision of TGCs in infancy like ours is much easier than in adults because of less inflammatory response^[4].

Conclusion

TGCs are embryological remnants of the hindgut and are lined with different epithelial types normally found in the gastrointestinal tract including stratified squamous, ciliated columnar, mucin-secreting columnar and transitional. It is important to differentiate it from other lesions in retrorectal space especially sacrococcygeal teratoma. Nevertheless, the possibility of tailgut cyst should be considered in the differential diagnosis of any retrorectal/presacral mass.

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