INDEX COPERNICUS INTERNATIONAL

Tailgut cyst and a very rare case of a tailgut cyst with mucinous adenocarcinoma in a 73 year old woman treated for buttock abscess with fistula

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Article history: Received: 12.02.2019 Accepted: 05.03.2019 Published: 07.03.2019

ABSTRACT: We present a case of malignant transformation of a tailgut cyst of retrorectal space in a 73-year-old woman. At the age of three she was treated for abscess and fistula of the right buttock, which periodically recurred. In 2016 she reported to a surgical outpatient clinic in Gorlitz with a large abscess of the right buttock and a purulent fistula. An abscess with a well-demarcated wall as well as the fistula were removed surgically. The cyst contained over a liter of purulent discharge. After it healed, a mucus fistula formed in the scar tissue. Histological examination showed a hamartoma retrocecal cyst with cells typical for mucinous adenocarcinoma. Magnetic resonance imaging was performed during her stay at the hospital, revealing residual lesion in the presacral region extending to S3 vertebra. Radical resection of the lesion was performed and confirmed with histopathological examination. No recurrence was noted during subsequent follow-up.

KEYWORDS: tailgut cyst, magnetic resonance imaging, buttock abscess with fistula

BACKGROUND:

Retrorectal cyst, or a tailgut cyst, is a rare congenital lesion presenting as a mass in the presacral space. It exists in the literature under many names: caudal intestinal cyst or distal intestinal cyst. English literature refers to it as myoepithelial rectal cystic hamartoma, rectal cyst, tailgut vestiges cyst, postnatal intestinal myoepithelial hamartoma, mucosecreting cyst, or enterogenous cyst. Tailgut cyst develops from residual posterior remnant of the intestine and is lined by varying glandular or transitional epithelium. It is a thin-walled, most often multi-chamber, branching structure. It rarely undergoes malignant transformation, which is related to a mutation of a gene responsible for inhibiting transformation of the p53 gene [1]. The distal part of intestine, from which the rectum and anus are formed, undergoes regression in the eight week of fetal life. The ectoderm invaginates into the end of intestine and anus is formed. However, disruption of this process may take place. Remnant of a stray fragment of the primitive gut, whose tail regresses, forms a tailgut cyst. Coccygeal fovea (fovea coccygea) located below the tailbone is most likely the result of filium terminale acting on the skin, pulling the anus toward the skin during embryogenesis [2]. Retrorectal space is limited anteriorly by the perirectal fascia – mesorectum, and posteriorly by the presecral fascia that covers the sacral bone. In simple terms: this space is demarcated anteriorly by the rectum and by the sacral bone posteriorly. Superiorly the roof of this space is formed by the peritoneal curvature and proper sacral fascia and presacral fascia. Inferiorly, the space is limited by muscles: levator ani muscle and coccygeal muscles. Levator ani is fixated by the rectococcygeal ligament to the coccygeal bone. Coccygeal muscles enforce it at the attachment site. They are covered by a strong fascia of pelvic diaphragm, which constitutes a part of the parietal fascia of the pelvis. Laterally, there are strands of pelvic fascia, ligaments and loose tissues, ureters and iliac vessels [3, 4].

Tailgut cysts are rare in children and extremely rare in adults. They occur more frequently in women (3:1) and the diagnosis is usually established between the age of 40 and 60. Cysts are usually diagnosed incidentally and may be associated with a broad spectrum of symptoms and complications. They are asymptomatic in about half of the cases. Symptoms are often associated with tumor mass or infectious complications and may present as fistulas. Other pathologies may also occur in this space. In children and newborns the most common lesions include teratomas (40% of tumors). It is not uncommon for teratomas to be very large and exceed 10 cm in diameter; in 10% of the cases they coexist with developmental disorders of body midline, such as encephalocele or spina bifida. Teratomas may be benign or malignant.

The majority of benign teratomas of the sacrococcygeal region are diagnosed in infants up to four months of age, while malignant tumors are more common in older children. Sacrococcygeal teratoma originates from 2 or 3 germ layers. Mature benign teratomas consist of cells that form tissues similar to normal (skin, hair, teeth, bone, intestine); vascular plexuses responsible for production of fluid are also frequently encountered. They may be demonstrated in a fetus as early as in the 1st trimester of pregnancy. In fetal life they occur with a frequency of 1:40 000 and are more common in females (4:1). Risk of malignant transformation reaches 40–50%. In 15% of cases lesions are cystic and in 85% – solid, which are usually malignant. Teratomas may develop upward or downward. Four types of sacrococcygeal teratomas can be distinguished based on this criterion according to the American Academy of Pediatric Surgical Section (AAPSS) classification:

- Most of the lesion is on the outside and only a small part is inside the body;
- Most of the lesion is on the outside with a notable portion inside the body;
- Most of the lesion is within the body and only a part is outside;
- Entire lesion is inside the body.

Types I / II constitute 80% and type IV – 10% of cases. Teratomas should be treated at reference centers for pediatric surgery. The

DOI: 10.5604/01.3001.0013.0810

Fig. 1. Extrarectal, presacral space with a pathological lesion.

above-mentioned classification is also useful in adult surgery for determining surgical access during removal of cysts located in the sacrorectal region [5, 6, 7, 8].

CASE REPORT:

Patient K.G., aged 73 years, reported that in childhood (at the age of 3) she was treated for an exudating fistula of the right buttock. For all of her adult life she has been suffering from purulent discharge from the fistula, for which she was treated conservatively. From the surgical outpatient clinic she was referred to hospital with a diagnosis of right-sided anal abscess and associated fistula with purulent exudate. She was admitted to the Surgical Ward of the Gorlitz Hospital on May 5, 2016. During this hospital stay the abscess and the fistula were removed surgically together with about 1 liter of foul-smelling pus.

After the wounds have healed a fistula recurred in the scar and green, mucinous discharge was present. Another surgery was unsuccessful. Histological examination of the excised lesion revealed retrocecal cyst hamartoma and mucinous adenocarcinoma cells. Magnetic resonance imaging of the pelvis was performed during the next admission to hospital. A lesion located superiorly and anteriorly to the coccyx reaching up to the level of S3 vertebra was identified. It was described as a complex of irregular cystic lesions spread over an area of 56x32x46 cm with a no-signal strand corresponding to a fistula in the surgical scar that extended toward the surface of the right buttock. Radical resection of the lesion was performed through inferior access below the coccyx, which was subsequently confirmed in histopathology. The wound healed successfully and the fistula did not recur.

Magnetic resonance (description: dr. Dariusz Szczerba): Figures 1.–4.

Magnetic resonance imaging demonstrated a complex of irregular cystic lesions spread over an area of 56x32x46 mm inferiorly and anteriorly to S3 vertebra and a no-signal strand corresponding to a fistula, surgical scar? that extended toward the skin of the right buttock and a 16-mm cyst was visualized on the edge of this strand, in the medial part of the buttock near gluteal sulcus. The above-described cystic structures surrounded by adjacent no-signal fibrous changes that distort the cysts – signs of retraction.

Cystic contents mostly resembled water, some cysts with increased signal in T1-weighted sequences, suggesting high protein content. Slight enhancement was noted along previously described fibers after contrast administration – scar tissue, inflammatory granulation?, and segmental enhancement along thickened walls of several cystic spaces. Beside that no contrast-enhancing masses suggestive of an ongoing neoplastic process were demonstrated.

Histological examination (description: dr. Andrzej Guzik): Figures 1.–3.

Surgical sample consisting of four tissue fragments, with total size 9x8x2 cm, collected from variously shaped cystic spaces was subject to histopathological examination. Walls of those spaces were made of fibrous connective tissue and some smooth muscle fibers, but contained no nerve plexuses. Individual spaces were lined with six types of epithelia, including bilayer and multilayer stratified cuboidal epithelium, ciliary epithelium, multilayer stratified keratinized epithelium, transitional epithelium and adenomatous epithelium (Fig. 1.). Some of those spaces were lined by one type of epithelium only, others by several types of epithelia. There were foci of lymphocyte- and histocyte-rich inflammatory infiltrates as well as pools of extracellular mucus in the cystic walls. Low-grade and high-grade dysplasia (Fig. 2a.) was found in some of the cystic spaces lined with adenomatous epithelium and one of them contained weaving of mucinous adenocarcinoma (Fig. 2b. and 2c.) developing over a 1.5-cm area. Destructive invasion of connective tissue stroma of cystic wall by mucinous adenocarcinoma was seen. There were, however, no neoplastic emboli within lymphatic and blood vessels.

Immunohistochemical staining of smooth muscle fibers found in cystic walls was SMA-positive (Fig. 3a.), some epithelia exhibited positive expression of cytokeratin 7 (ciliary and transitional epithelium) (Fig. 3b.) and some were positive for cytokeratin 20 (adenomatous epithelium of colon type) (Fig. 3c.). It is worth noting that almost 100% of mucinous adenocarcinoma cells developing in one of the tailgut cysts overexpressed the p53 protein (Fig. 3d.) had a mitotic index over 50% and stained positive for Ki-67 antibody. Dysplastic adenomatous epithelium exhibited moderate expression of p53 protein and Ki-67 antibody staining. On the other hand, there was no p53 expression in the benign adenomatous epithelium and mitotic index of the Ki-67 antibody remained < 2%. The whole picture indicates rapid proliferation in the malignant adenomatous epithelium and p53 overexpression suggests that malignant transformation within the tailgut cyst resulted from p53 gene mutation [1].

DISCUSSION

Primary retrorectal tumors include: neurogenic tumors, osteogenic tumors, metastases, and presacral meningocele. Congenital tumors constitute 55–65% of all tumors in this region, neurogenic tumors 10–12%, osteogenic tumors 5–11%, inflammatory tumors 5% and other types 5–11%. According to their embryonic origin cysts are classified into: epidermal, dermal, neural, teratomas, en-

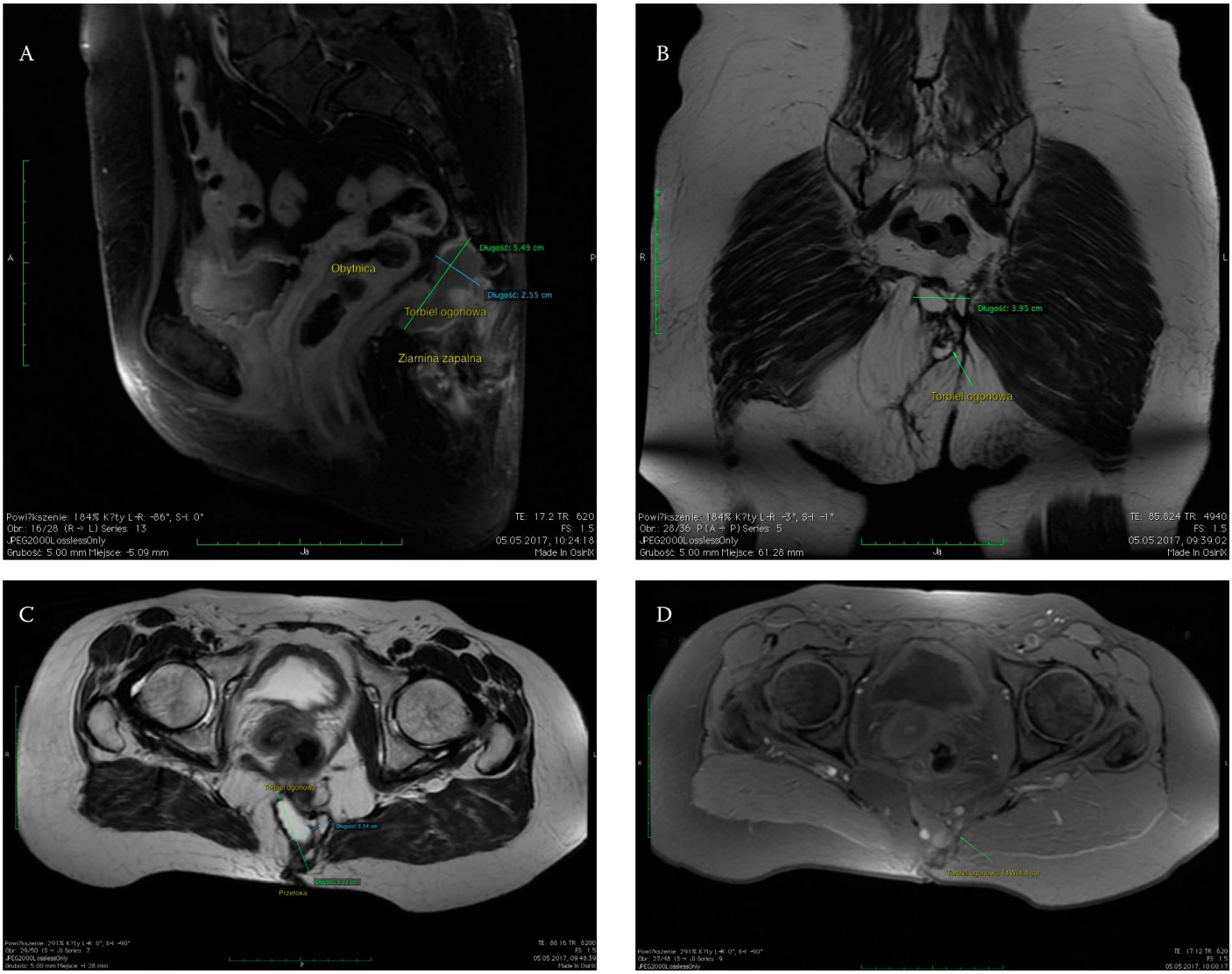


Fig. 2. MR images.

teric cyst, rectal duplication cyst, mucous secreting cyst, enterogenous cyst, simplex cyst, gland anal cyst, rectal cyst, hamartoma, tailgut cyst.Epidermal cyst is usually unbranched, consists of a single chamber and is lined with multilayer squamous epithelium. It originates from a protrusion of epidermal elements during closure of meningeal groove. Dermal cyst is branched and contains elements such as: skin, apocrine glands, sebaceous glands, hair follicles, tooth buds; it communicates with the skin. Inflammatory cyst may manifest as slit-shaped ulcer. Gland anal cyst may contain epithelium similar to that in the tailgut cyst, but occurs in a different location (rectal mucus membrane) and absence of smooth muscle germ cells. Tailgut cyst consists of three elements: ectodermal, endodermal and mesodermal. Cyst epithelium may very: mucinous or ciliary columnar, pseudostratified, stratified squamous, transitional cuboidal, mucus-secreting cuboidal, transitional, or columnar. Tailgut cyst is thin-walled, may be single- or multi-chamber (a conglomerate of cysts), branched, benign or malignant, may contain green opalescent colloid fluid. Rectal duplication cyst is lined with epithelium similar to that of gastrointestinal or respiratory tract with crypts. Mucus membrane of this type of cyst resembles intestinal. Cyst has well-demarcated walls made of smooth muscle cells and contains nerve plexuses [9, 10, 11]. Tail-gut cyst is often asymptomatic or presents with atypical symptoms. Thus, the diagnosis is often made late. It is frequently diagnosed incidentally. Presenting symptoms result from the mass of the tumor: pain in the perineum, anus, lesser pelvis, during defecation, changed stool thickness, bleeding, difficulty defecating, dysuria, or even urinary retention. Purulent fistula may be the first symptom and may be identified as anorectal fistula, recurrent perianal abscess or pilonidal cyst. Thorough per rectum examination or gynecological examination may enable detection of the tumor on the posterior wall of the rectum, although it is not always palpable. In case of purulent perianal fistula tailgut cyst may be easily mistaken for pilonidal cyst or even anal abscess and fistula, as in the above-described case, or with an anorectal fistula. Transrectal ultrasound, computed tomography and especially magnetic resonance are useful for establishing proper diagnosis.

Radical surgical excision of the cyst is the first-line treatment. It is particularly important due to the possibility of malignant transformation as in the described case. According to other authors posterior surgical access is preferred if the lesion is located below S3 level (as in this case) and coccygeal bone can be removed if necessary. Inserting finger into the anus makes dissection of this area easier. Tissues in this region are not easy to dissect, while vessels and nerves are hidden from view. When a lesion is large and extends above S3 then laparotomy is indicated. Depending on the

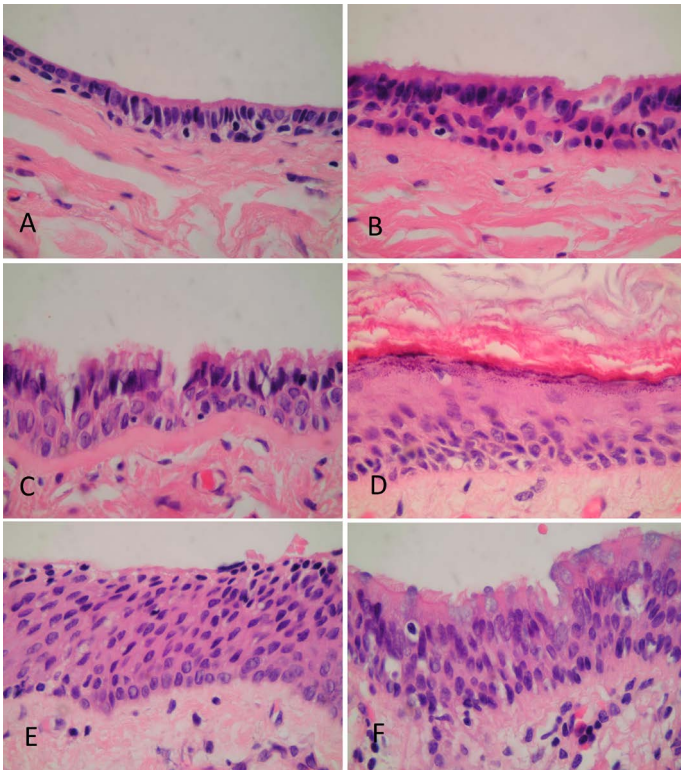
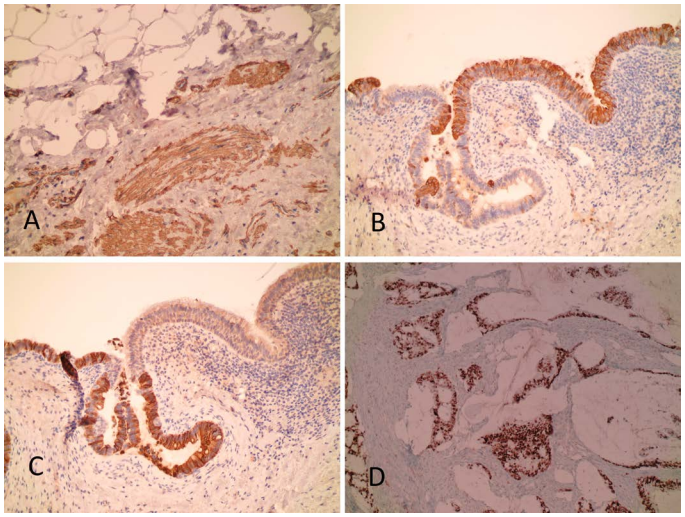


Fig. 3. Types of epithelia lining the walls of the tailgut cyst: (A) bilayer cuboidal epithelium (H+E; zoom 100x); (B) multilayer cuboidal epithelium (H+E; zoom 100x); (C) ciliary epithelium (H+E; zoom 100x), (D) multilayer squamous keratinized epithelium (H+E; zoom 100x), (E) transitional epithelium (H+E; zoom 100x), (F) adenomatous epithelium (H+E; zoom 100x).



Ryc. 5. Immunohistochemical staining: (A) SMA-positive smooth muscle fibers (zoom 20x); (B) positive expression of cytokeratin 7 in ciliary and transitional epithelium (zoom 20x); (C) positive expression of cytokeratin 20 in colon-type adenomatous epithelium (zoom 20x); (D) overexpression of p53 protein in almost 100% of cells of mucinous adenocarcinoma developing in the tailgut cyst (zoom 20x).

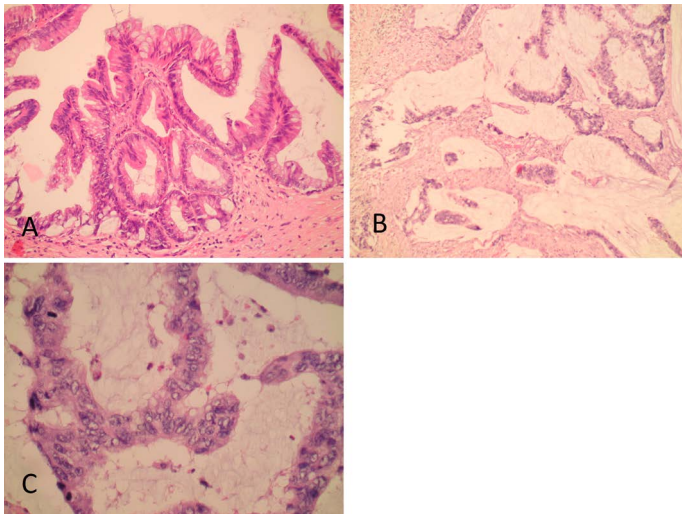


Fig. 4. (A) Low-grade dysplasia of the adenomatous epithelium lining some of the cystic spaces of the tailgut cyst (H+E; zoom 20x); (B) mucinous adenocarcinoma in the tailgut cyst (H+E; zoom 20x); (C) mucinous adenocarcinoma in the tailgut cyst (H+E; zoom 40x).

location and size of a lesion other authors used the following access routes: transrectal, transperineal, transsacral, transvaginal, transsphincteric, transcoccygeal, or laparoscopic. Puncture is indicated in case of a large cyst containing a lot of fluid [12, 13, 14].

Scarce reports found in international literature reflect rare incidence of tailgut cysts. Case series of patients gathered throughout several dozen years come from large clinical centers. Cysts that have undergone malignant transformation are particularly rare. Middeldorpf was the first to describe a tailgut cyst in 1885. Other authors described: 53 cases in patients aged 4 days to 73 years, 43 cases gathered over 16 years, 30 cases collected over a period of 30 years, 16 cases of a tailgut cyst undergoing malignant transformation and another 12 cases (5 – carcinoid, 7 – adenocarcinoma); single-case descriptions include women aged 29, 46, 49, 64 and 68 years and men aged 19, 34 and 72 years [12, 13, 14, 15, 16, 17, 18, 19]. We did not encounter any descriptions of a tail-gut cyst in an adult person in Polish literature. In the presented case it is particularly notable that the described cyst has been symptomatic since birth, was associated with purulent fistula on the buttock and underwent malignant transformation.

CONCLUSIONS

Malignant transformation of a tailgut cyst of retrorectal space in an elderly woman is extremely rare. It can be taken for an abscess and fistula of the buttock. MRI is a very important examination, which allowed for a diagnosis in the presented case. The basic treatment method is radical resection of the lesion.

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Word count: 2870 Page count: 5 Tables: – Figures: 5 References: 19

DOI: 10.5604/01.3001.0013.0810 Table of content: https://ppch.pl/issue/11974

Competing interests: The authors declare that they have no competing interests.

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Cite this article as: Rachel F., Guzik A., Szczerba D., Kozieł K., Gutterch K.: Tailgut cyst and a very rare case of a tailgut cyst with mucinous adenocarcinoma in a 73-year-old woman treated for buttock abscess with a fistular; Pol Przegl Chir 2019: 91 (3): 38–42