Unusual cause of CA 19.9 elevation diagnosed by endoscopic ultrasound-guided fine needle aspiration: a retrorectal tailgut cyst

A 42-year-old woman underwent a magnetic resonance imaging (MRI) scan for CA 19.9 elevation, noted during a routine evaluation. Endoscopic ultrasound-guided fine needle aspiration (EUS-FNA) of a retrorectal cystic mass of unknown etiology was scheduled. A 5-cm multiseptated cystic lesion was noted (Fig. 1). Under antibiotic prophylaxis, FNA was performed (19-gauge needle; single pass); the aspirate was thick and yellowish. Cytological examination showed squamous epithelial cells but no atypia (Fig. 2). In-house MRI confirmed a multicystic lesion with a hyperintense signal on T1-weighted images (Fig. 3). The patient opted for a laparoscopic resection, which confirmed a tailgut cyst (Fig. 4), with later CA 19.9 normalization.

The human embryo possesses a true tail as an extension of the primitive gut. A retrorectal cystic hamartoma (tailgut cyst) is a rare congenital lesion representing a nonregressed tail. Usually found in asymptomatic middle-aged women, local mass effect or complications, namely malignant degeneration, have been described [1]. Ultrasound shows multilocular cystic lesions with internal echoes due to mucoid material or inflammatory debris. Surgical excision is the gold-standard treatment, with the laparoscopic approach being the most recent option [2]. There is only one report of EUS-FNA with a flexible echoendoscope [3]. Puncture should be performed when other etiologies are considered or if malignant degeneration changes management. Finally, clinicians should be aware that benign tailgut cysts are a rare cause of mild CA 19-9 elevation [4].

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Fig. 1 Endoscopic ultrasound (EUS) image: retrorectal cystic lesion with echogenic foci. Detail: additional small cystic lesion adjacent to the anal canal, which has been described as a potential cause of recurrence.

Fig. 2 Fine needle aspiration (FNA) smears showing proteinaceous background, squamous cells without atypia, anucleated squames, microcalcifications, and debris (Papanicolaou stain, ×100).

Fig. 3 Multicystic retrorectal lesion. Several cysts with a high signal in a T1-weighted image, interspersed in a fibrous septa, reported in mucus fluid with high protein concentration.
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Fig. 4 Multilocular cystic lesion with squamous, transitional, and glandular epithelial lining; mucous and keratin contents (Hematoxylin and eosin, original magnification ×100).