Presacral tumors of the Currarino triad: teratomas or hamartomas?

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Purpose The aim was to elucidate the nature of the presacral tumors in the Currarino triad through studying their preoperative radiological anatomy and histopathology of excised specimens.

Patients and methods The study group included three operated cases of Currarino triad. All were women who presented with constipation and demonstrated the typical three components of the disease: anorectal anomaly, sacral bony defect, and presacral tumor. The histopathological slides of excised specimens (presacral tumors) were available for re-examination. For comparison, we included another 'control' group representing the standard sacrococcygeal teratomas (without vertebral or anorectal anomalies).

Results Histopathological examination of presacral tumors in the Currarino triad showed multicystic spaces lined by different types of epithelia mainly keratinized stratified squamous epithelium with focal areas of transitional epithelium. The underlying stroma showed fibrovascular connective tissue admixed with randomly arranged smooth muscle bundles. In contrast to the standard sacrococcygeal teratomas, neither skin adnexal structures nor heterologous mesenchymal tissues were observed; no immature elements could be detected.

Conclusion In the Currarino triad, several clinical and histopathological observations would suggest the excised presacral tumors to be developmental cysts (retrorectal hamartomas) rather than neoplastic teratomas. *Ann Pediatr Surg* 14:137–142 © 2018 Annals of Pediatric Surgery.

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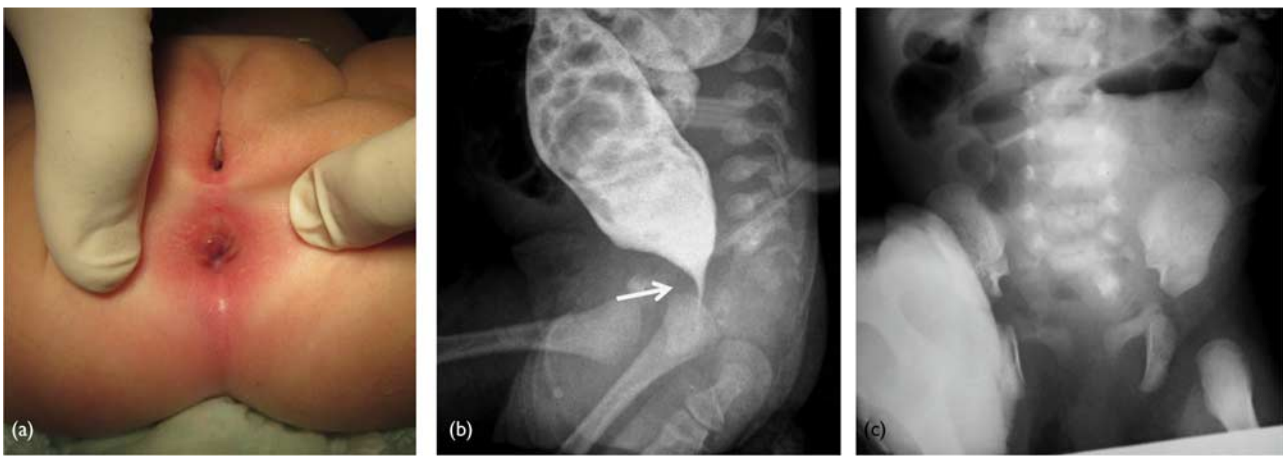
Introduction

Currarino triad is a rare congenital disorder characterized by the presence of three components: anorectal anomaly, sacral bony defect, and a presacral mass [1,2]. The latter may be either a lipomyelomeningocele, or a tumor commonly described in the literature as presacral teratoma [3,4].

The presacral tumors associating the Currrarino triad tend to behave differently from other sacrococcygeal teratomas. Dirix *et al.* [5] have shown the former to be associated with a very low risk of malignant transformation compared with the latter. Moreover, Weinberg [6] suggested that the term 'teratoma' in Currarino triad was

Fig. 1



Case 1: a 12-month-old female with the Currarino triad: anorectal stenosis, sacral bony defect, and presacral mass. (a) Normal external appearance: the presacral mass has no external component, and the anal canal is normally positioned but stenotic 'funnel anus'. (b) Contrast enema showing characteristic anorectal stenosis (arrow). (c) Plain radiograph anteroposterior view showing the characteristic notched sacrum 'scimitar' indicating for the presacral mass.

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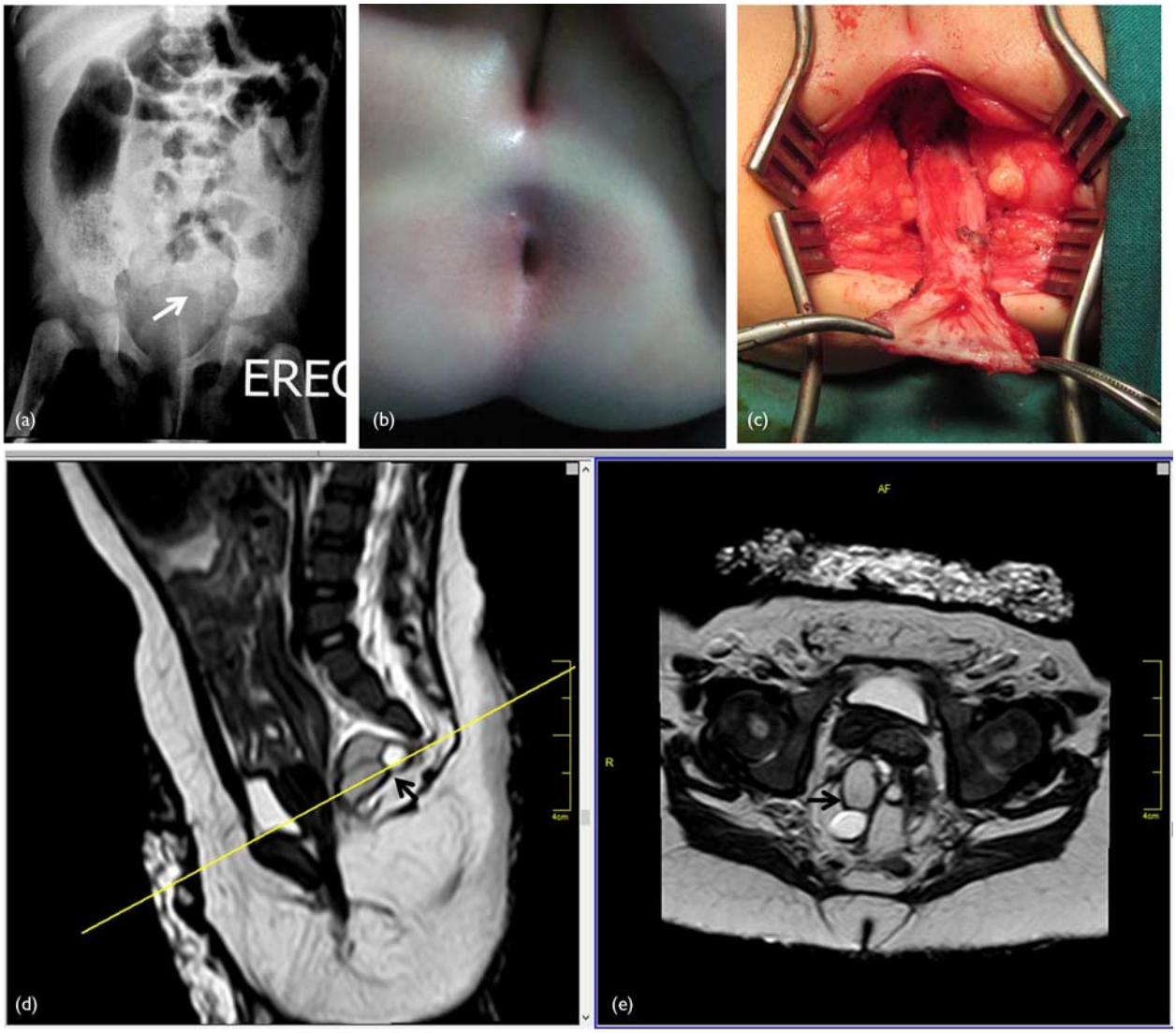
Fig. 2



Case 2: an 8-month-old female with the Currarino triad: anorectal stenosis, sacral bony defect, and presacral mass. (a) Plain radiograph anteroposterior view showing the characteristic notched sacrum 'scimitar' (white arrow) indicating for the presacral mass. (b) Normal external appearance: the presacral mass has no external component, and the anal canal is normally positioned but stenotic 'funnel anus'. (c) Anorectoplasty with excision of the stenotic distal anal canal. (d, e) MRI T2WI (midsagittal and axial cuts) demonstrating the presacral multicystic mass (black arrow), exhibiting variable signal intensity that reflects the variable protein content of the fluid inside. The straight yellow line in (d) is marking for the level of the axial plane in (e). Note the sacral deformity (incomplete sacrum) in the sagittal plane.

just a 'misnomer', and that the correct pathological terminology should be hamartoma.

In this report, we tried to elucidate the nature of the presacral tumors in the Currarino triad through studying their preoperative radiological anatomy and the histology of excised specimens, while comparing the results with another group of standard sacrococcygeal teratomas (without vertebral or anorectal anomalies).

Patients and methods

During the period 2010–2016, 17 cases of Currarino triad were diagnosed and treated at our Pediatric Surgery Department [7]. Among these cases, six were associated with presacral tumors that were excised and subjected to histopathological examination. The medical records of these patients were retrospectively inspected for clinical data, preoperative investigations, operative details, and pathological reports of excised specimens (all had a pathological diagnosis of mature cystic teratoma). In this study, we included cases whose histopathological slides of excised specimens were available and could be retrieved for re-examination.

For comparison, we included another 'control' group representing the standard sacrococcygeal teratomas. The study was conducted after internal review board approval.

Results

The study group included three operated cases of the Currarino triad. All were female patients who presented with constipation and demonstrated the typical three components of the disease: anorectal anomaly, sacral bony defect (notched sacrum or 'scimitar' deformity), and presacral mass. Their clinical and radiological findings

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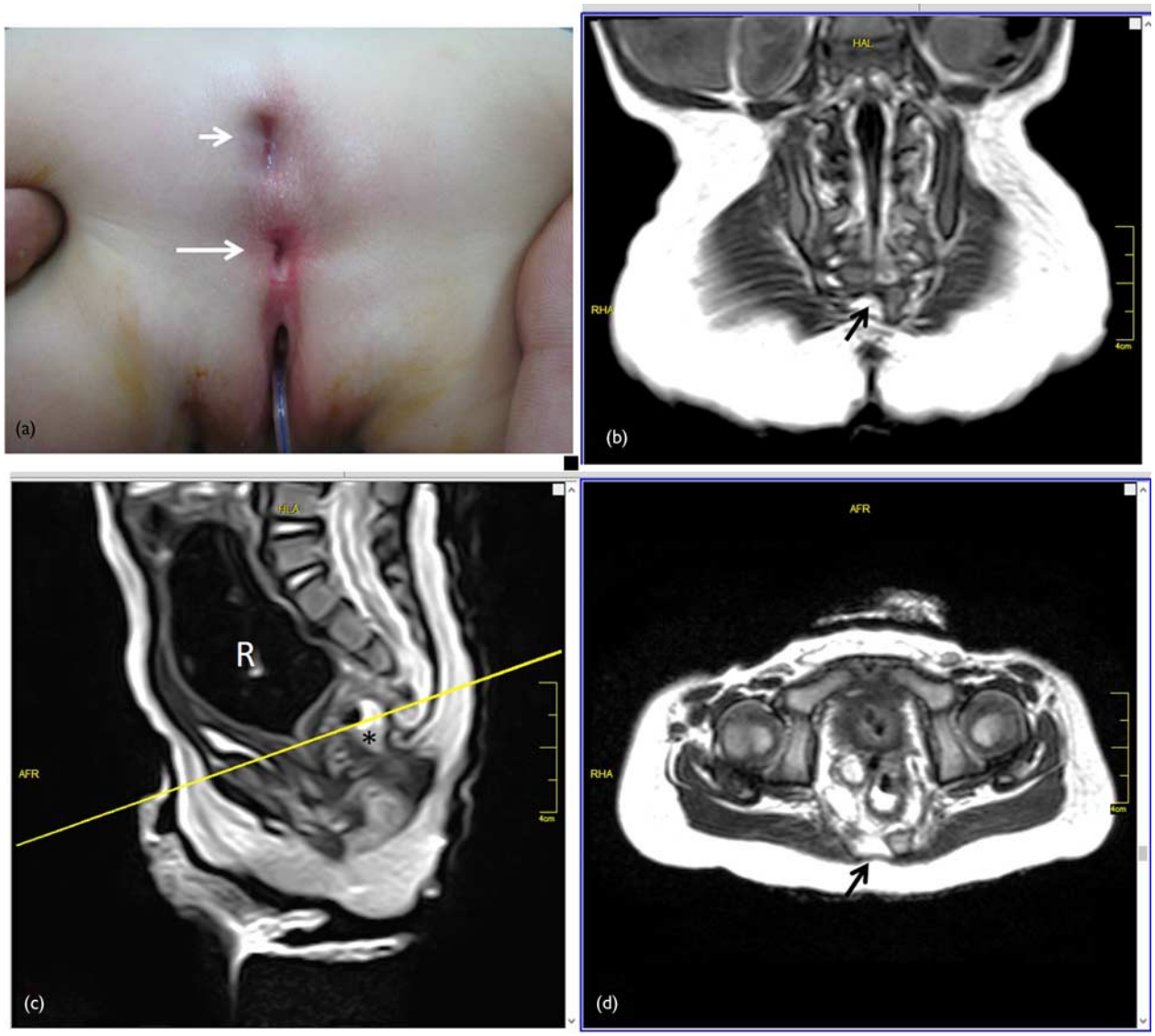
Fig. 3



Case 3: a 24-month-old female with the Currarino triad: anorectal anomaly, sacral bony defect, and presacral mass. (a) The patient is in the prone position; notice the anteriorly displaced and stenotic anus (long white arrow), and the postanal sinus (short white arrow) resulting from infected presacral cyst. (b) MRI (coronal section) showing a small sacral defect 'notch' (black arrow). (c) MRI (T2WI, midsagittal plane) demonstrating the presacral mass (asterisk). Note the distended rectum (R). The straight yellow line is marking for the level of the axial plane in (d). (d) MRI (T2WI, axial plane) demonstrating the multicystic nature of the presacral mass. The black arrow is pointing to the hemivertebral defect of the distal sacrum.

are summarized in Figs 1–3. The three cases underwent surgery for correction of anorectal anomalies (anterior ectopic anus/anorectal stenosis) and excision of presacral tumors. Their histopathological slides were available for re-examination. In the three cases, histopathological examination showed multicystic spaces lined by different types of epithelia mainly keratinized stratified squamous epithelium with focal areas of transitional epithelium (Fig. 4). Keratin flakes were seen within some cystic spaces. The underlying stroma showed fibrovascular connective tissue admixed with randomly arranged smooth muscle bundles. Neither skin adnexal structures nor heterogeneous mesenchymal tissues (bone, cartilage) were observed. No immature elements could be detected.

The control group included four female cases of operated sacrococcygeal teratomas, whose pathological slides were available for comparison (Fig. 5). Although the size of sacrococcygeal teratomas was variable ranging from small (< 5 cm) to large (>10 cm) tumors, characteristically there were no associations with other developmental anomalies. MRI studies demonstrated complete sacrum and coccyx (Fig. 6) and a normally developed anorectum that was compressed by the mass effect of the tumor. Another clinical difference from presacral tumors found in the Currarino triad was the constant presence of an external component of the mass related to the coccyx in cases of sacrococcygeal teratomas (Fig. 6).

Discussion

Developmental 'presacral' or 'retrorectal' cysts are quite rare. However, their significance lies in the frequently encountered diagnostic difficulties, liability for complications, and the risk

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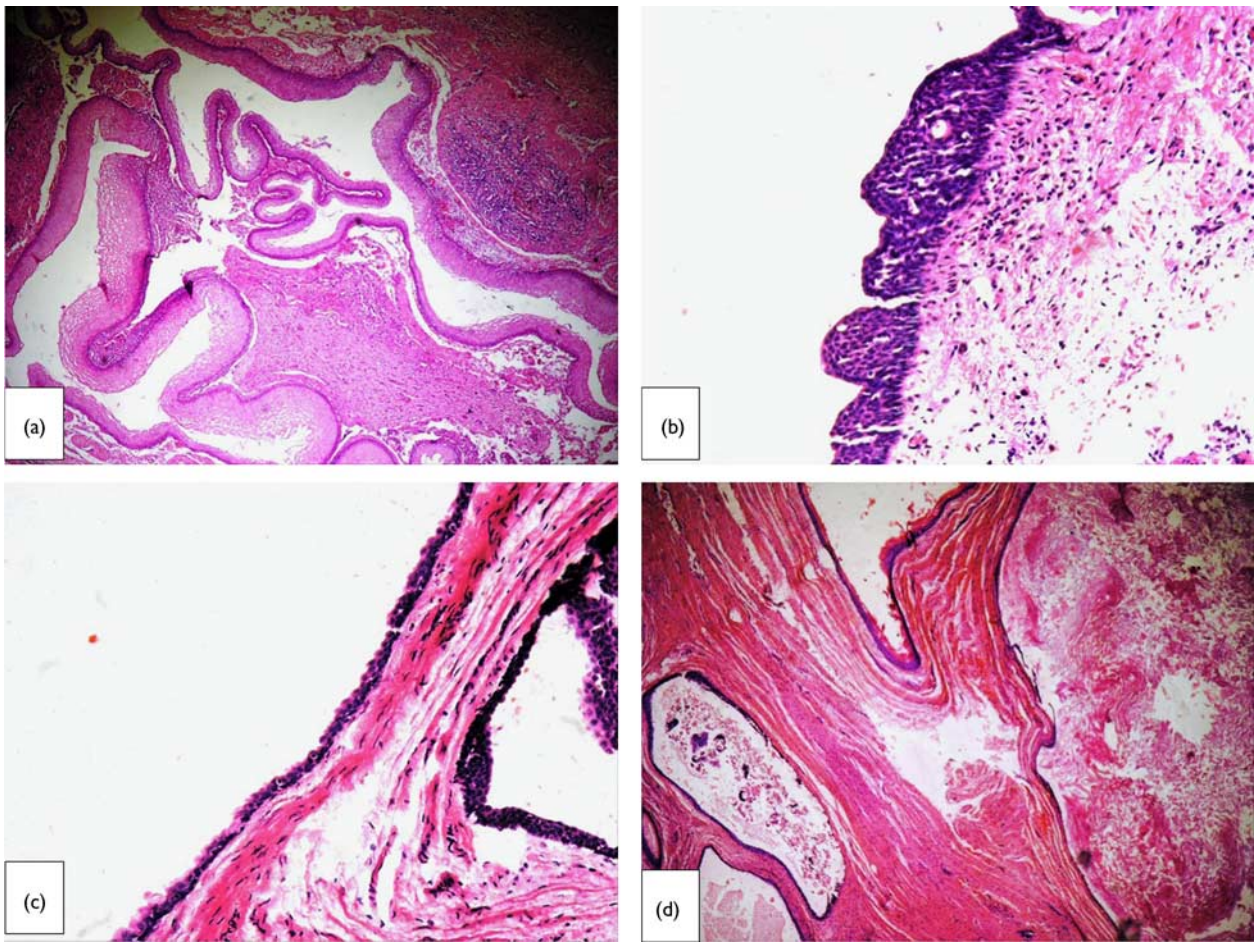
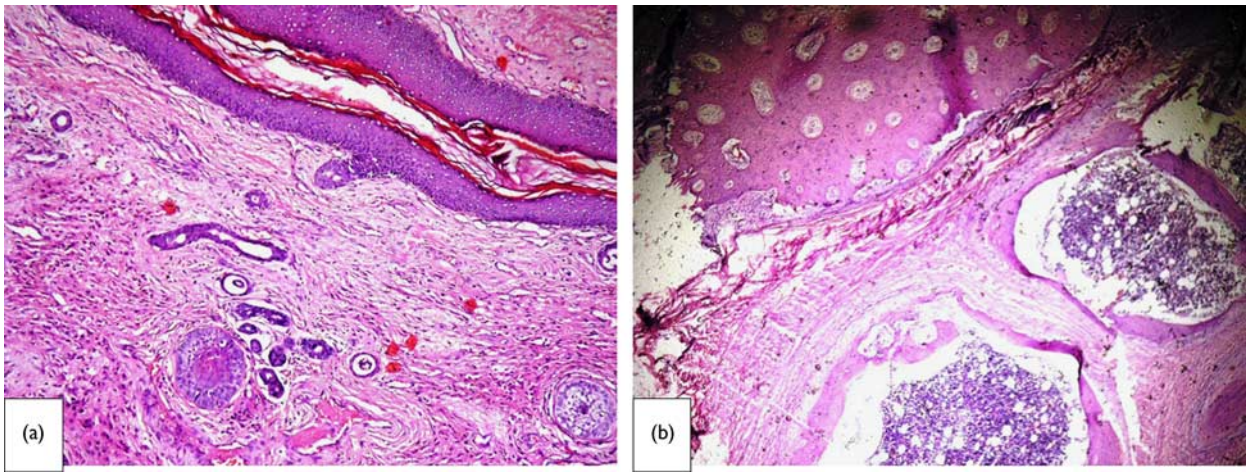
of malignant transformation [8]. On the basis of differentiating histopathological features, the developmental presacral cysts can be classified into dermoid, epidermoid, and enteric cysts [9]. The latter can be further subclassified into either rectal duplication or tailgut cysts (also known as retrorectal hamartomas) [10]. Other differential diagnoses

Fig. 4



Histopathological examination of the 'Currarino triad' cases showed multicystic spaces lined by different types of epithelia comprising. (a) Keratinized stratified squamous; (b) transitional; (c) simple pseudostratified; and (d) keratin flakes were seen within some cystic spaces. Neither skin adenexal structures nor heterologous mesenchymal tissues were detected (hematoxylin and eosin; a and d: original magnification ×40, b and c: original magnification ×200).

Fig. 5



Histopathological examination of operated sacrococcygeal teratomas cases showed (a) cystic space lined by keratinized stratified squamous epithelium with the underlying skin adenexal structures and (b) heterologous mesenchymal tissue in the form of bone tissue (hematoxylin and eosin; original magnification ×100).

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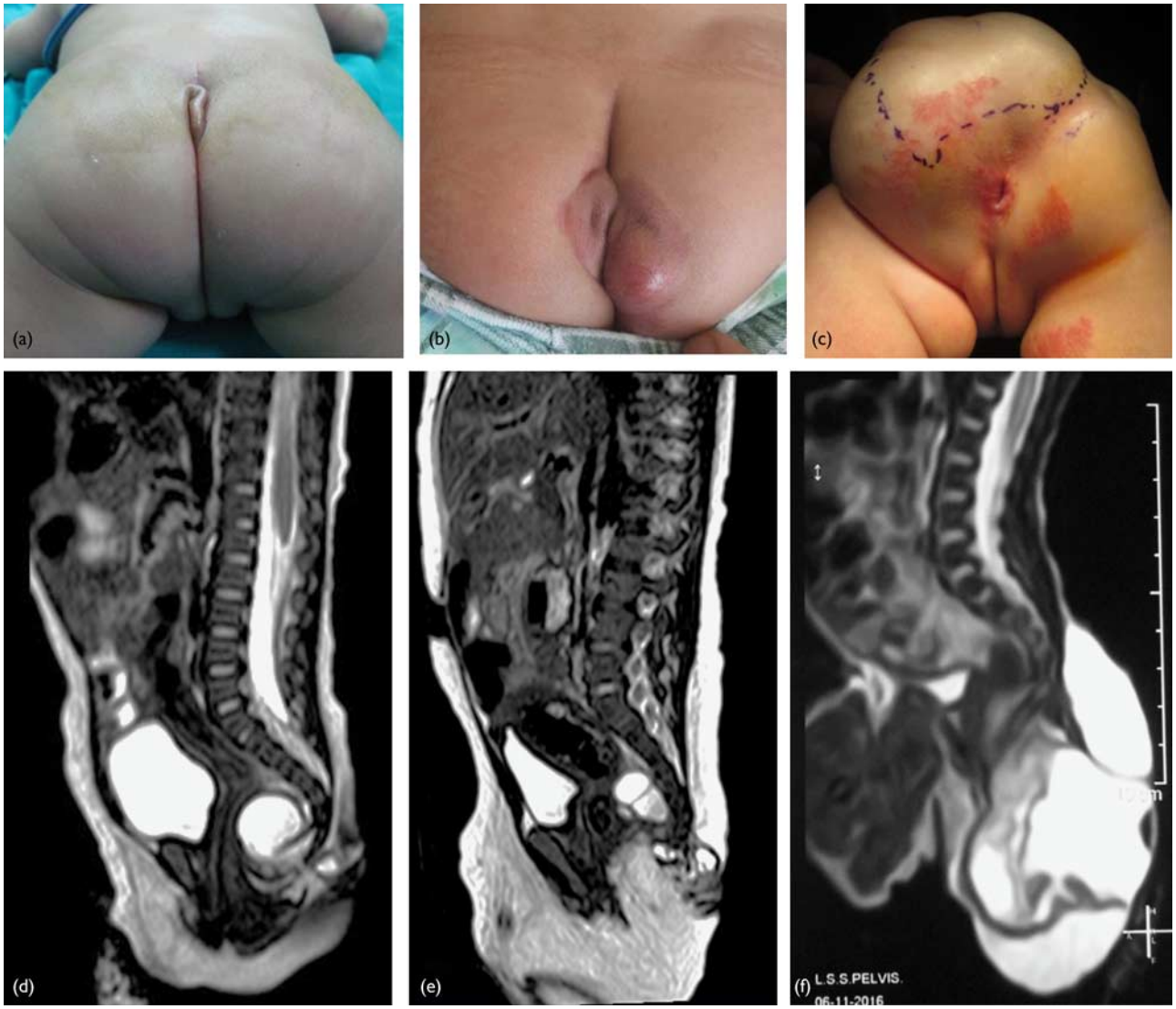
Fig. 6



Three female cases with sacrococcygeal teratomas (the control group). The upper row (a, b, c) the external appearance of the three cases; always there was an apparent external component that ranged from a very small component in (a) to a large one (c), and all had a normally developed anus. The lower row (d, e, f) represents the corresponding midsagittal MRI of the three cases in the upper row. Note the completeness of the sacrum and coccyx in the control group in contrast to the Currarino triad.

of cystic lesions in this area include cystic teratomas, shwannomas, and anterior meningoceles [11].

Presacral tumors associating the Currarino triad have been frequently described in the literature as presacral teratomas [1,5]. Moreover, it might have been included in Altman's classification as sacrococcygeal teratoma 'type IV' [12]. However, we have several observations (pathological, clinical, embryological, and radiological) that would highly suggest these tumors to be retrorectal hamartomas (tailgut cysts) rather than neoplastic teratomas.

Most studies would rely on histopathological features to differentiate between various types of presacral cystic lesions. The presence of different types of epithelial lining within tailgut cysts (retrorectal hamartomas) in the pediatric age group might have been a source of confusion with the totipotent teratomas (the most common tumor in the neonatal period); even some authors advocated that there was no clear distinction between these two pathological entities [9]. However, the main criterion to distinguish a teratoma is to demonstrate cells originating from the three germ layers [9] or presence of immature components, which were not found in any of our cases of the Currarino triad. In 1988, Hjermstad and Helwig [13] were the first to provide a detailed pathological description of tailgut cysts in a large series of 53 cases (predominantly women) [13]. They referred to the diagnostic importance of finding transitional or glandular epithelium among the variety of epithelia present in tailgut cysts. In our study group, the focal areas of transitional epithelium were found in all studied cases (three cases). The presence of scattered 'disorganized' smooth muscle fibers is another histopathological feature of tailgut cysts [9,10,14], which was also demonstrated in the revised slides of our study group. This has been described as a differentiating feature of the tailgut from rectal duplication cysts (the latter have a well-developed smooth muscle layer) [9,10,14].

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Clinically, the presentation of presacral tumors in the Currarino triad is very similar to what has been described in the literature concerning tailgut cysts (retrorectal hamartomas) except for the delayed presentation in the latter (typically middle-aged women). Similarities include the characteristic female predominance, strict presacral location of the mass with normal external appearance, superadded infection (pelvic abscess, recurrent perianal fistula), constipation, and the low incidence of malignant transformation [5,7,14,15]. The presence of anorectal anomalies in the Currarino triad may be responsible for early recognition of the disease through aggravating constipation [7]. The very infrequent reporting on vertebral and/or anorectal defects among cases of retrorectal hamartomas in adults [16] might be explained by the presence of an incomplete form of the disease or by the presence of very minor defects that could have been easily overlooked [7].

The well-recognized constellation of congenital anomalies (vertebral and anorectal) in the Currarino triad can indicate for the developmental nature of presacral cysts in these cases. On the contrary, the neoplastic sacrococcygeal teratomas (although frequently reaching a huge size) are associated with a complete sacrum and coccyx, and a normally developed (but compressed) anorectum. Tailgut cysts (retrorectal hamartomas) are thought to be derived from remnants of the embryonic tailgut (primitive hindgut), or remnants of the neurenteric canal [11,17]. The embryonic tailgut, which is located between the cloaca anteriorly and the notocord posteriorly represents that part of the gut distal to the developing anal canal. By the sixth week of gestation, the embryonic tailgut should have normally regressed via a process of programmed cell death (apoptosis), which takes place in a craniocaudal direction [18]. It has been suggested that aberrations in the process of tailgut regression would hamper the normal development and differentiation of the anorectum from the cloaca and result in anorectal anomalies [19]. Similarly, its proximity to the notocord (which appears inseparable from the embryonic tailgut distally) [18] can provide an explanation for the distal vertebral bony defect present in these cases.

Diagnosis of tailgut cysts in adults by different imaging modalities is frequently challenging with a long list of differential diagnoses [11,20]. Typically, tailgut cysts appear as retrorectal (presacral) cystic masses, frequently multilocular, and with variable signal intensities in MRI (depending on the protein content of the fluid inside the cysts) [11,21]. Cysts complicated with superadded infection demonstrate strong cyst wall enhancement. Again, these imaging features were quite like what we have found in cases of the Currarino triad [7]. Adding this imaging similarity to the above-mentioned similarities (histopathological, clinical, and embryological explanation), we believe that the presacral cystic tumors in the Currarino triad represent a variety of tailgut cysts. The presence of associated anorectal anomalies and vertebral defects may be responsible for their earlier recognition and presentation in the pediatric age group.

Conclusion

In the Currarino triad, several clinical and histopathological observations would suggest the excised presacral tumors to be developmental cysts (retrorectal hamartomas) rather than neoplastic teratomas.

Conflicts of interest

There are no conflicts of interest.

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