CASE REPORT

Atypical presentation of sacrococcygeal yolk sac tumor in infant: beware of the injuries of the gluteal region

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Abstract Malignant sacrococcygeal yolk sac tumor is a rare extra-gonadal germ cell tumor of infancy and childhood. We report the case of a 14-month-old white female infant with a rapidly growing supra-gluteal mass at first misdiagnosed as hemangioma after a clinical assessment. The lesion was then classified as extra-gonadal yolk sac tumor due to alarming ultrasound features, later confirmed at MRI and pathology. This report remarks the need of a rigorous methodology in the ultrasound exploration of the gluteal region.

Keywords Yolk sac tumor · Germ cell tumor · Sacrococcygeal region · Ultrasound

Riassunto Lo Yolk sac è un raro tumore della linea germinale dell'infanzia e dell'adolescenza. Riportiamo il caso di una piccola paziente di 1 anno e 2 mesi, giunta alla nostra osservazione ecografica per il riscontro clinico di una tumefazione palpabile, riferita in progressiva crescita, in corrispondenza della regione glutea, erroneamente ipotizzata essere un angioma. La lesione mostrava caratteristiche ecostrutturali sospette per patologia a carattere evolutivo - infiltrativo come rilevato anche alla Risonanza Magnetica ed alla successiva indagine istopatologica con diagnosi definitiva di Yolk sac Tumor. La nostra esperienza

sottolinea la necessità di un approccio rigoroso nell'indagine ecografica nella valutazione della regione glutea.

Introduction

Sacrococcygeal yolk sac tumor (YST), also known as “endodermal sinus tumor”, is a malignancy of infancy and childhood of rare occurrence. Here, we describe the case of a primary YST presenting as a supra-gluteal soft mass and its ultrasound appearance warranting urgent surgical intervention.

Description of the case

A 14-month-old, Caucasian female infant came to our attention for a midline supra-gluteal mass grown over the previous 2 weeks. The infant presented as a healthy baby with timely achievement of the appropriate neurodevelopmental milestones and no significant medical history. The only abnormality at her physical exam was a soft, oval, 2 by 3 cm mass covered by normal-looking skin. The lesion was labeled as hemangioma by the primary care physician because it was compressible and not adherent to the deeper structures.

Ultrasound (US) exploration was started with the patient in prone position on the posterior midline using a 12–15-MHz frequency probe (Aloka SSD-5000, Wallingford, CT, USA).

Dynamic gray-scale sonography of the subcutaneous tissue showed a large oval mass measuring $32 \times 19 \times 36$ mm with sharp margins and a solid component was predominant with mixed echogenic–hypoechoic

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Fig. 1 Gray-scale longitudinal sonogram in prone patient position with high-frequency probe shows a well-defined oval mass with sharp margins and predominant solid component with inhomogeneous mixed echogenic-hypoechoic echotexture

Fig. 2 Color Doppler longitudinal sonogram in prone patient position with high-frequency probe shows a well-defined, slightly hypoechoic mass with increased peripheral and internal flow

echotexture (Fig. 1). Color Doppler imaging demonstrated a very intense blood flow both around and within the lesion (Fig. 2). Pulsed Doppler investigation of the arteries supplying the lesion detected high velocity and low resistance suggesting a hemangiomatic mass. This color Doppler finding, detected with linear high-frequency probe, seemed to confirm the clinical hypothesis. However, the presence of deep vascular poles at color Doppler prompted us to complete the examination of the patient in prone and in supine position using a 5-MHz frequency probe with the same scanner. This precaution led to the detection of a considerably larger lesion ($54 \times 36 \times 50$ mm) deepening from the sacrum to dislocate the bladder to the right (Fig. 3). Since hemangiomas are very uncommon in the

Fig. 3 Gray-scale longitudinal sonogram in supine patient position with low-frequency probe shows the mass (M) compresses the bladder (B) on the right

Fig. 4 Pelvic MRI axial T1-w TSE mdc mod: bulky mixed pelvic tumor (black arrow) compresses the bladder on the right (white arrowhead)

deep pelvis, any mass/tumor occurring in this region mandated to rule out a malignancy.

The subsequent diagnostic program included a pelvic magnetic resonance imaging (MRI) scan that confirmed a large heterogeneous pelvic mass extending from the gluteal region to include the coccyx extending deep into the pelvis with a considerable mass effect (Fig. 4). An incisional biopsy was done to get a tissue sample that had a histological and immunohistochemical pattern consistent with a yolk sac tumor. A total body computerized tomography (CT) scan was also performed to complete the tumor staging; no liver or lung metastases were found and retroperitoneal lymph nodes were not enlarged. The patient received chemotherapy for tumor reduction and then surgery with mass and coccyx resection in toto. At the time of the diagnosis, serum alpha-fetoprotein marker was found to be

greater than 2,500 ng/mL (normal range 0.6–11.1 ng/mL) and it dramatically decreased after surgery. The patient continues to do well, currently followed as an outpatient by our pediatric oncologists.

Discussion

First described by Teilum in 1959 as “endodermal sinus tumor”, YST is a teratoid neoplasm with selective differentiation toward vitelline (yolk sac) structures [1]. YST is generally classified among germ cell tumors (GCTs), a heterogeneous group also including teratoma, choriocarcinoma, germinoma, and embryonal carcinoma [1, 2]. The two most common histologic patterns are a pure yolk sac tumor and another associated with teratoma, dermoid cyst, or choriocarcinoma [3]. Malignant germ cell tumors collectively account for only 3 % of pediatric malignancies and occur more often in girls than in boys [2]. GCTs tend to present on the midline or paraxial sites of the body and can be gonadal or extra-gonadal. The latter is an unusual occurrence, mostly in the mediastinum, the pineal gland, the vagina or, like in our case, the sacrococcygeal region [2, 3]. Altman suggested a clinical classification of sacrococcygeal tumors with a type I (46 %), external lesion with low malignant potential to an uncommon and highly aggressive type IV (10 %), situated deep into the pelvis or the abdomen [4, 5]. Infants and young children who present with a presacral mass often develop abdominal, urinary, or rectal complaints or even a low back and sacral pain. A differential diagnosis should exclude masses of the presacral area in their congenital, developmental or inflammatory origin [2, 5].

Imaging techniques should be used in combination to define with precision the location and the extension of presacral masses of early infancy and childhood [5].

Sonography plays a crucial role in this respect rapidly conveying accurate information with no need for ionizing radiations.

The ultrasound exploration of the patient in both supine and prone position turned out to be critical in avoiding the original hemangioma misdiagnosis.

Because of its rarity, there has not been a detailed description of the ultrasound features of YST. However, the ultrasound characteristics of GCTs depend on their contents [5]. GCTs may be solid, cystic, or a combination of both [2]. At US, a yolk sac tumor is a mostly

hyperechoic solid mass, though it may contain cystic regions [3]. A predominant cystic component with occasional calcifications is a feature of a mature benign sacrococcygeal teratoma, whereas a mostly solid lesion, like in our case, is suggestive of malignancy. Hemorrhage and necrosis are common [5]. Color Doppler interrogation in some tumors may show marked hypervascularity and look like a hemangioma: the location of the mass, however, should raise the need for further investigation. Malignant teratomas may metastasize or extend into adjacent structures such as the spine or gluteal muscle [5], as in our case. However, sonographic features are poorly predictive of malignancy though the patient’s age and the elevated alpha-fetoprotein are valuable clues [5].

In conclusion, this case highlights the value of ultrasound for the initial evaluation of pediatric patients with gluteal soft mass. We stress the concept that in early infancy a gluteal mass can actually represent the tip of an “iceberg” invading deep into anterior structures. We thus firmly recommend to scan these lesions with a proper combination of low and high-frequency probes in both the prone and the supine position.

Conflict of interest Francesco Esposito, Marco Di Serafino, Patrizia Oresta declare that they have no conflict of interest.

Informed consent All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, and its late amendments. Additional informed consent was obtained from all patients for which identifying information is not included in this article.

Human and animal rights This article does not contain any studies with human or animal subjects performed by any of the authors.

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