Monophasic Synovial Sarcoma Presenting as Infantile Hemangioma: A Case Report

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Case report

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Abstract

Objective: This report describes a case of a monophasic synovial sarcoma that was initially misdiagnosed as an infantile hemangioma in a 3-month-old female infant. We also discuss the basic elements of evaluating a soft-tissue mass and important radiological and clinical features that should raise the index of suspicion for a synovial sarcoma.

Summary Background Data: Synovial sarcoma is a rare pathology that is often challenging to diagnose due to its similarities with more common vascular lesions that occur in infancy. Infantile hemangioma, in particular, is common, has distinctive clinical and radiological findings, and is frequently diagnosed without histological confirmation. This increases the risk of misdiagnosis of rare tumors in infancy.

Case Report: A 3-month-old female infant presented with a soft-tissue mass on the left forearm. Physical examination showed a well-defined, subcutaneous mass on the left forearm, measuring approximately 3 cm × 5 cm, firm in consistency, with no cutaneous involvement and normal skin coverage. Imaging suggested an infantile hemangioma but, due to the atypical presentation, a trial of beta-blocker therapy was conducted to confirm the diagnosis. The patient showed no clinical response to beta-blocker therapy, and therefore an incisional biopsy was performed. Using histopathology, the tumor was diagnosed as a monophasic synovial sarcoma.

Conclusions: We highlight a rare presentation of a synovial sarcoma in a neonate and caution against making a diagnosis without histopathologic analysis. Clinicians should include sarcoma in the differential diagnosis when assessing a mass in neonates, as early diagnosis and intervention can reduce morbidity and mortality.

MINI ABSTRACT A 3-month-old infant presented with a left forearm soft-tissue mass, initially diagnosed as infantile hemangioma. Biopsy revealed a monophasic synovial sarcoma. We discuss the basic elements of evaluating a soft-tissue mass and important radiological and clinical features that should raise the index of suspicion for a synovial sarcoma.

1. Introduction

Misdiagnosis of a malignant tumor, such as a synovial sarcoma, as a benign tumor, such as a hemangioma, is disastrous, as malignant tumors have poor prognoses. Both benign and malignant soft-tissue tumors characteristically present as a painless, growing prominence [1]. Many physicians have a misconception that there is no need to worry unless a mass is painful, when, in reality, most malignant tumors are asymptomatic early in the course of the disease [1]. In addition, the absence of constitutional symptoms such as fever and weight loss should not lower the physician's index of suspicion of a malignancy when evaluating a soft-tissue mass, as these symptoms rarely present even in the case of metastasis [2]. In particular, neither a synovial sarcoma nor infantile hemangioma (IH) present with constitutional symptoms. A slowly growing soft-tissue mass typically indicates a benign lesion, whereas a rapidly growing soft-tissue mass (growing over a period of weeks to months) may indicate the

malignant nature of a lesion [1]. The waxing and waning size of a soft-tissue mass is not a characteristic feature of a sarcoma; this may indicate a ganglion cyst or hemangioma [1]. Currently, polymerase chain reaction (PCR) is the gold standard for the diagnosis of a synovial sarcoma [3]. However, IH is diagnosed clinically based on the presence of raspberry-red cutaneous stains, requiring neither imaging studies nor biopsy. Because a synovial sarcoma can have an unusual presentation, resulting in delayed diagnosis and management, and is a relatively rare entity, it must be in the differential diagnosis when assessing a neonate with a palpable mass, as it may otherwise be misdiagnosed as a more common benign tumor such as an IH. We present a case of a monophasic synovial sarcoma in a 3-month old infant that was initially misdiagnosed as a hemangioma.

2. Case Report

Consent to publish this case report was obtained. This report does not contain any personal information that could lead to the identification of the patient.

A 3-month-old female infant was referred to our center for the evaluation of a mass on the left forearm, which had been present for 2 months. The infant was otherwise healthy with an unremarkable medical history. The parents first noticed a mass on the infant's left forearm at the age of 1 month. The parents reported that the mass was stable in size, had no color changes, had normal surrounding skin, and no bleeding or discharge. On admission, the infant was conscious and alert with no signs of distress. On examination, there was a well-defined, subcutaneous mass on the left forearm, measuring approximately 3 cm × 5 cm, firm in consistency, with no cutaneous involvement, and normal skin coverage (Fig. 1). Initial laboratory test results were unremarkable. Ultrasound (US) and MRI suggested an IH with intramuscular extension, but clear separation from the muscle tissue could not be observed using MRI. Due to the atypical presentation of the mass, a trial of beta-blocker therapy was conducted to confirm the diagnosis. No clinical response was observed, and there was even a question of a possible increase in the size of the mass. Consent to perform surgery was obtained. An urgent histopathological examination was performed using an incisional biopsy. This showed features of a monophasic synovial sarcoma. The child then travelled abroad with her parents for further management and follow-up.

3. Discussion

3.1 Differentiating benign and malignant soft-tissue masses

Indicators to differentiate an IH from other soft-tissue tumors have been described by Frieden [4]. Features that can assist the clinician in the evaluation of a soft-tissue mass include the size, depth, consistency, and mobility of the mass. Benign masses tend to be superficial and smaller than 5 cm in diameter. In contrast, malignant lesions are typically found deep to the fascia and are larger than 5 cm in diameter. However, sarcomas are superficial in approximately 30% of cases. The growth characteristics of the tumor are important to consider [4]. Knight considered onset during the neonatal period to be an important indicator of an IH, as onset and accelerated growth during the first few weeks of life are a sine

qua non of the condition [5]. However, an IH rarely presents as a fully formed tumor at birth. This presentation would narrow the differential diagnosis down to a congenital hemangioma, rare vascular tumor, or other malignant soft-tissue tumors [4]. In this case, the mass was noticed by the parents during the first month of life. Steady growth of a tumor that continues beyond 6–9 months could suggest other diagnoses, as most IHs stabilize in size by this age [4]. Another clue to the diagnosis was the fixation of the mass to the fascia [4]. Fixation to the fascia is uncommon in IHs, as most IHs and congenital hemangiomas are found within the dermis and subcutaneous fat [4]. Another sign was the firm consistency of the mass [5]. On palpation, the mass, in this case, was found to have an unusually firm consistency. IHs and congenital hemangiomas can be firm initially; although, this can be subjective [5]. Soft and mobile masses are likely to be benign tumors such as lipoma, whereas malignant lesions tend to be firm and fixed [1]. Lipomas are the most common small, superficial masses in adults, while hemangiomas are the most common in children [1].

3.2 Imaging of IHs

The diagnosis of an IH is clinical, but imaging is indicated in cases of uncertain diagnosis, critical anatomic locations, or complications [6]. Imaging methods for evaluating a soft-tissue mass include plain radiography, computed tomography (CT), and MRI. The echogenicity of a hemangioma varies depending on its soft-tissue and vascular components, which vary throughout its normal life cycle [6]. During progression, the lesion can be more hypoechogenic, while increasing echogenicity can be observed during regression. High diastolic velocity, high vessel density, and a Doppler shift of more than 2 kHz can be observed using Doppler US [7].

3.3 Imaging of synovial sarcomas

Synovial sarcomas should be evaluated using imaging modalities [8]. Primary lesions and distant metastases can be detected by radiological investigation; although, images may appear normal in up to 50% of cases if the lesion is small [9]. The first imaging modality used should be plain radiography, which may reveal any bony involvement [8, 13]. In addition, the presence of calcifications can be assessed using plain radiography; these are present in approximately 30% of sarcoma cases [10]. A soft-tissue mass, periosteal reaction, and bone invasion are other possible radiographic features, which indicate a synovial sarcoma [8] rather than an IH [11]. US can be used to differentiate solid and cystic masses [8], and it has a high negative-predictive value for soft-tissue masses. Our patient presented at the age of 3 months with a radiological diagnosis of a hemangioma, following US and MRI evaluation. US revealed a large, well-defined, subcutaneous hypoechoic lesion, with echogenic foci indicating calcifications, on the medial aspect of the left forearm, measuring 44 mm × 22 mm (Figure 2). Using color Doppler, the lesion appeared markedly vascular.

The role of CT in diagnosing a soft-tissue mass has declined with the use of MRI. However, CT can be used in patients who cannot undergo MRI, and it can be useful in differentiating calcifications from ossifications. Distant metastases can also be detected using CT [12]. MRI is the imaging modality of choice for assessing a soft-tissue mass in a child [8, 13]. MRI can show the typical features of a synovial

sarcoma, including a lesion smaller than 5 cm in diameter, with regular margins and diffuse heterogeneous patterns of enhancement [14]. In our case, MRI revealed a well-defined mass lesion measuring 30 mm × 23 mm × 52 mm, occupying the anterior and medial aspects of the forearm from the elbow to the wrist, with blood vessels traversing it. The mass could not be separated from the muscle compartments in the same region and was not related to the bone.

3.4 Histopathologic analysis

Biopsy should be performed on all suspicious masses to reach a diagnosis [8]. In this case, the lesion was unusually hard and deep-seated, making it highly suspicious for malignancy. Furthermore, a poor clinical response was observed following propranolol (Inderal) administration for 2 weeks. A tissue biopsy was therefore performed. Soft-tissue masses can be evaluated by needle biopsy or open biopsy. Needle biopsy includes fine-needle aspiration (FNA) and core needle biopsy (CNB), while open biopsy is divided into incisional and excisional biopsies. US can be used intraoperatively to assist with the biopsy or excision, especially if the mass is not palpable. Histologically, there are three subtypes of synovial sarcomas: monophasic, biphasic, and poorly differentiated [12]. The most common subtype is the monophasic synovial sarcoma [15], which is comprised of spindle cells [12]. Biphasic synovial sarcomas have spindle-cell and epithelial-cell elements [12]. In this case, incisional biopsy revealed a monophasic synovial sarcoma pattern. Cytokeratins (CK) or epithelial membrane antigens (EMA) are expressed in over 90% of cases of synovial sarcomas [15]. However, they are minimally expressed in spindle cells and thus, can be very sparse [15]. In addition, spindle cell synovial sarcomas are mostly positive for BCL2, CD99, and calponin [15]. Therefore, synovial sarcomas can be identified by staining for a combination of CK, EMA, BCL2, CD99, and calponin [15]. Synovial sarcomas have similar histologic characteristics in children and adults [16, 17]. In this case, immunohistochemistry (IHC) revealed that the tumor was positive for TLE1, CD99, BCL2, and vimentin, but negative for desmin, SMA, CD34, synaptophysin, CK, and EMA. Histopathologic analysis revealed that the tumor cells had hyperchromatic nuclei with frequent mitosis and were invading the skeletal muscle and adjacent soft tissue.

3.5 Diagnosis of benign and malignant soft-tissue tumors

Due to the rarity of neonatal malignancies and the overlap in the presentation of benign and malignant soft-tissue tumors, sarcomas must be in the differential diagnosis when assessing a neonate with a palpable mass, as they may otherwise be misdiagnosed as more common benign tumors such as IHs. To the best of our knowledge, there has previously been a single case of a biphasic synovial sarcoma that was initially misdiagnosed as a hemangioma [15]. Review of the literature also revealed three cases of congenital infantile fibrosarcomas that were misdiagnosed as ulcerated IHs [18]. In this case, the onset soon after birth, persistent growth despite treatment with propranolol (Inderal), and the firm consistency of the mass led us to question the initial diagnosis of a hemangioma and obtain a tissue biopsy that led to the final diagnosis of a synovial sarcoma.

Conclusion

We report an unusual case of a monophasic synovial sarcoma with histopathologic confirmation, which mimicked a benign lesion, resulting in a delayed diagnosis. We also discuss the basic elements of evaluating a soft-tissue mass. We highlight a rare presentation of a synovial sarcoma in a neonate and caution against making a diagnosis without histopathologic analysis. Clinicians should include sarcoma in the differential diagnosis when assessing a mass in neonates, as early diagnosis and intervention can reduce morbidity and mortality.

Declarations

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Figures

Figure 1

Subcutaneous mass on the left forearm of a 3-month-old infant

Figure 2

Ultrasound image of a subcutaneous mass on the left forearm of a 3-month-old infant