An Unusual Presentation of a Congenital Myofibroma: A Case Report

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INTRODUCTION

Infantile Myofibroma (IM) is a rare benign mesenchymal soft tissue tumor manifested by a solitary nodular lesion involving the skin, subcutaneous tissues, muscles, bone or viscera. These lesions are composed of contractile myofibroblastic cells arranged around thin-walled blood vessels and primarily occur in neonates or infants, with few reports of adult onset. IM presents as firm, flesh- or purple-colored solitary or multiple nodules that can be sporadic, congenital or familial, with most lesions occurring in the head and neck region. Treatment is almost always successful with complete excision of the lesion. Although the prognosis for solitary IM lesions is excellent and recurrence is unusual, these tumors, when involving the visera, are associated with poor prognosis and can be fatal when involving the gastrointestinal or cardiorespiratory systems. It is important to consider together the clinical, histological and immunohistochemical features to make an accurate diagnosis and proceed with the proper treatment.

CASE DESCRIPTION

History of Present Illness: This is a 1 day old biracial (Asian/Caucasian) male who presented with a soft well-circumscribed flesh-colored left cheek mass with a prominent network of blood vessels at birth. On day 2, the lesion began to form a central area of necrosis. From Day 2 to 7, the mass became more firm. On Day 7, the mass was biopsied and because of the extensive necrosis, the lesion was completely excised.

Past Medical History: Mother, 32-year-old, gravida 2, para 2 at 39 weeks gestation. Delivered via cesarean section and with an unremarkable pregnancy. Weight, 8 lb 10 oz and Apgar scores of 9 and 10 at 1 and 5 minutes respectively.

Family History: Noncontributory.

Physical Exam: A 3.0 x 3.0 cm solitary, firm, dome shaped, well circumscribed, flesh-pink-colored dermal/subcutaneous nodule on the left zygomatic arch with a rich network of blood vessels and a central area necrosis. No facial asymmetry or structural facial defects were appreciated and no other lesions of the skin or deeper tissues were noted.

Imaging: Brain MRI without contrast revealed a well-circumscribed mass in the subcutaneous soft tissues of the left zygomatic arch measuring 1.3 cm transverse x 2.4 cm anteroposteriorly x 2.0 cm craniocaudal. The mass was slightly heterogeneous but predominantly T1 and T2 hypointense. No feeding vessels leading to the mass were noted. The mass appeared to be separated from the left orbit, and no intracranial extensions of the mass were noted. A small cystic structure within the temporal horn of the left lateral ventricle was noted. Otherwise, the ventricular system was normal in size, shape and configuration. No intracranial masses or midline shift were noted. The visualized major intracranial vessels appeared patent. No abnormality was seen in visualized portions of the orbits. The middle ear cavities, mastoid air cells and paranasal sinuses were clear.

Treatment: The patient underwent surgical excision. The post-operative course was uneventful and no recurrence was observed after long-term follow-up.

OBJECTIVE

The purpose of this case report is to present a unique clinical vignette and progression of the solitary form of infantile myofibroma.

HISTOPATHOLOGY AND IMMUNOHISTOCHEMISTRY

Histopathology: Sections showed normal skin with an underlying dermal and subcutaneous lesion demonstrating a peripheral rim of viable tissue with a necrotic center. The viable cells had oval nuclei with small, non-prominent nucleoli. They were associated with a rich network of vessels, some demonstrating a branching "hemangiopericytoma-like" pattern. Scattered mitotic figures were also noted.

Immunohistochemistry: Cells stained positive for smooth muscle actin (SMA). CD31 staining highlighted the numerous vessels with their focal branching pattern.

DISCUSSION

• The congenital infantile myofibroma is a very rare disorder, with less than a hundred documented cutaneous cases worldwide.
• Although first described as "congenital generalized fibromatosi" by Stout in 1954, it was not until 1981 when Chung and Enzinger coined the term "infantile myofibromatosi" based on age of onset and histologic characteristics of the tumor.
• Three clinical scenarios of IM are recognized and include solitary IM, multicentric IM without visceral involvement and multicentric IM with visceral involvement. The solitary IM is the most common form that presents as a single, firm, non-tender nodule ranging from 0.5 to 7 cm.
• Solitary lesions usually occur in the skin, muscles, bones, and soft tissues anywhere in the body. Solitary myofibromas of gastrointestinal tract, larynx and lungs, kidneys, ovaries, and testes have also been previously reported.
• Affects males more commonly than females
• More than one third of lesions occur in the head and neck region and 90% of the 61 cases, reviewed in one study, were diagnosed in patients younger than 2 years of age.
• Symptoms may vary depending on the location and size of the lesion. Reduced mobility of the hand, stridor, and flaccid weakness in the lower limb were reported in the literature with lesions involving brachial plexus, larynx and spinal cord respectively.
• Surgical removal of the tumor is the only definitive treatment and was a treatment of choice in the present case.
• As reported in several case reports/series, solitary myofibroma have a variety of clinical presentations.

CONCLUSION

This case illustrates a rare example of solitary cutaneous congenital myofibroma. While spontaneous regression of solitary lesions has been documented, surgical resection is the only definitive treatment. Given the rarity of this condition, a correct preoperative diagnosis is important and often is challenging to make. The differential diagnosis initially favored malignancy due to the area of necrosis. Therefore, it is important that the physician be familiar with this rare type of lesions when working up soft tissue lesions of infancy and childhood. Note: there was significant freeze artifact complicating the histological diagnosis of myofibroma. But in conjunction with the clinical picture and immunohistochemistry results, the diagnosis of myofibroma was made.

REFERENCES


TABLE 1. IMMUNOHISTOCHEMICAL PROFILE

<table>
<thead>
<tr>
<th>Antibody</th>
<th>Vimentin</th>
<th>α-SMA</th>
<th>S-100 protein</th>
<th>Desmin</th>
<th>CD34</th>
<th>Pan-cytokeratin</th>
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<tr>
<td>Result</td>
<td>+</td>
<td>+</td>
<td>(&lt; 5%)</td>
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TABLE 2. DIFFERENTIAL

<table>
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<tr>
<th>CONDITION</th>
<th>FEATURES</th>
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<tr>
<td>Infantile Hemangioma</td>
<td>• Most common tumor of infancy</td>
</tr>
<tr>
<td>Infantile Fibrosarcoma</td>
<td>• Typically appear between 1 and 4 weeks</td>
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<tr>
<td>Rhabdomyosarcoma</td>
<td>• Mesenchymal cell tumor that is composed of malignant fibroblasts in a collagen background</td>
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<tr>
<td>Leiomysarcoma</td>
<td>• Present in the first 2 years of life</td>
</tr>
<tr>
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<td></td>
</tr>
<tr>
<td>Desmin (++)</td>
<td></td>
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<tr>
<td>Keratin (++)</td>
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Figure 2. (A) Hemangiopericytoma-like pattern in the dermis. (B) Viable tissue with necrotic center.

Figure 1. Infantile Solitary Myofibroma (A) at the time of delivery. (B) at 2 days old, and (C) at 6 days old.

DISCUSSION