

Solitary Infantile Myofibromatosis; Differential Diagnosis of Solitary Frontal Bone Lytic Lesion in Neonatal Period

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Abstract

Background: Infantile myofibromatosis (IM) is the most common benign fibrous soft tissue tumor found in infants, presenting either as a single lesion (solitary) or multiple lesions (multicentric) that can affect the skin, muscles, bones, or internal organs. While bone involvement is typically associated with the multicentric form or craniofacial bones in solitary cases, it rarely appears in the extremities, where it radiologically presents as a well-defined, bone-dissolving lesion surrounded by a hardened border. **Case Presentation:** A 6-month-old male infant with a congenital left frontal bone swelling. Radiological evaluation revealed a well-circumscribed, expansile lytic skull lesion with no internal extension or abdominal masses. Complete surgical excision and subsequent microscopic and immunohistochemical examination confirmed the benign diagnosis, showing a characteristic biphasic pattern of SMA-positive, spindle-shaped cells and vascular spaces without any signs of malignancy. **Conclusion:** By combining clinical observations and imaging with characteristic biphasic microscopic features, solitary infantile myofibromatosis can be accurately identified and differentiated from similar spindle cell tumors, ensuring a positive outlook for the patient.

Keywords: Infantile Myofibromatosis- Solitary Infantile Myofibromatosis- Osteolytic Lesion- Benign Spindle Cell Lesion

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Introduction

Infantile myofibromatosis (IM) is the most common fibrous neoplasm of infancy. It is a disorder characterized by the overgrowth of soft tissue within the skin, muscles, bones, or visceral organs. It was first described by Stout in 1954 and was originally termed congenital generalized fibromatosis [1]. The lesion was then renamed “myofibromatosis” by Chung and Enzinger in 1981 [2] to reflect its myofibroblastic component. The World Health Organization (WHO) classification of soft tissue tumors in 2002 categorized IM under the category of benign fibroblastic/myofibroblastic lesions.

There are two distinct clinical presentations of IM: solitary and multicentric forms, the latter of which may have visceral involvement [3]. Solitary infantile myofibromatosis (SIM) tends to occur more frequently in males within the first two years of life, usually affecting the skin, subcutaneous tissues, or deep soft tissues. Osseous involvement by IM is more common in the multicentric variant, and most cases of osseous involvement by SIM

affect craniofacial bones. Few reports in the literature have described osseous involvement by SIM in the extremities [4]. Radiologically, osseous SIM appears as a well-circumscribed osteolytic lesion surrounded by a sclerosing margin [5].

Case Presentation

A 6-month-old male infant was referred to the Neurosurgery Department at Sohag University Hospital because of a solitary left frontal swelling present since birth. The infant did not exhibit any syndromic features or dysmorphic facies. There were no other surface body swellings or skin pigmentation, and there was no family history of similar conditions.

Brain MRI showed a well-circumscribed, expansile, lytic lesion involving the left side of the frontal bone, causing thinning of the outer and inner tables. The lesion measured approximately $2.5 \times 1 \times 2.5$ cm in maximum dimensions and exhibited low signal intensity on

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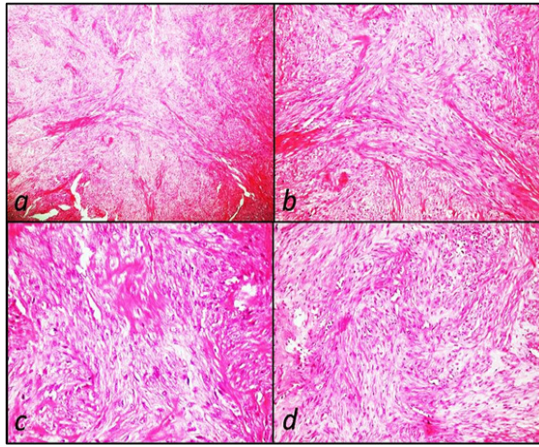


Figure 1. Microscopic Features of Excised Frontal Bone Mass in A 6-month-old Male Infant (H&E stain). (a; $\times 40$): Demonstrates the characteristic multinodular and biphasic architecture of infantile myofibromatosis. (b; $\times 100$): Shows the interface between the two distinct zones. Bundles and fascicles of well-differentiated, bland spindle-shaped myofibroblasts are seen merging into dense, collagenous stroma. (c & d; $\times 200$): Reveal the detailed cellular morphology. The hypercellular areas are composed of uniform, short spindle cells featuring bland, elongated nuclei and a moderate-to-abundant amount of eosinophilic cytoplasm. Interstitial thin-walled, branching, and slit-like vascular spaces are scattered throughout the stroma. Notably, there is a complete absence of cellular atypia, nuclear pleomorphism, necrosis, or atypical mitotic figures, confirming the benign nature of the lesion.

T1-weighted images and moderately low signal intensity on T2-weighted images. No evidence of intradural extension was found. An abdominal ultrasound did not reveal any masses.

The lesion was excised, and the specimen was sent to the Pathology Laboratory at Sohag University Hospital. The specimen was received as multiple, greyish-white, firm tissue fragments, collectively measuring about 4×3.5 cm. Microscopic evaluation revealed a biphasic lesion composed of central, hyalinized, hypocellular, collagenous areas surrounded by hypercellular areas. The latter were composed of bland-looking, short, spindle-shaped cells. These cells had short spindle nuclei with abundant eosinophilic cytoplasm. The stroma showed thin-walled, branched, and slit-shaped vascular spaces. No nuclear pleomorphism or mitotic figures were detected. Thorough examination of the tissue sections didn't reveal eosinophils, lymphocytes, multinucleated giant cells, or extravasated erythrocytes (Figure 1). Immunohistochemically; the proliferated spindle cells showed diffuse and strong expression for SMA (Figure 2). Based on the clinical, radiological, microscopic and immunohistochemical features, a diagnosis of solitary infantile myofibromatosis was made.

Discussion

Infantile myofibromatosis (IM) is a rare, benign mesenchymal disorder characterized by fibroblastic/myofibroblastic proliferation. It represents the most

common fibrous tumor in infancy and young children, with an incidence ranging from 1 in 150,000 to 400,000 live births [1]. Approximately 90% of IM cases appear within the first two years of life [6]. Immunohistochemically, the tumor cells express smooth muscle actin (SMA) and vimentin, while they are negative for desmin, S100, epithelial membrane antigen (EMA), and cytokeratin [7].

IM can be solitary or multicentric, and differentiation between these two variants depends on clinical and radiological evaluation. Solitary infantile myofibromatosis (SIM) is the most frequent and least aggressive variant [1]. One-third of SIM cases involve the head and neck, while the upper extremities are the second most frequently involved sites [4]. The prognosis of IM depends on the presence or absence of visceral involvement; SIM has a favorable prognosis, and some cases have shown spontaneous regression. In contrast, visceral involvement in multicentric infantile myofibromatosis is associated with high mortality rates because of gastrointestinal, cardiac, pulmonary, or central nervous system involvement [3].

In the current case, we report on a 6-month-old male infant who presented with a left frontal bone osteolytic lesion. The mass was excised, and histopathological evaluation revealed hyalinized fibrocollagenous bands surrounded by proliferating, bland-looking, spindle-shaped cells. The cells possessed short spindle nuclei and eosinophilic cytoplasm. No mitotic figures or pleomorphic features were detected. The main differential diagnosis for this case included neurofibroma, nodular fasciitis, hemangiopericytoma, leiomyoma, and fibrosarcoma.

Neurofibroma is a rare peripheral nerve sheath tumor. Microscopically, it is composed of a mixture of haphazardly arranged, wavy spindle cells, Schwann cells, and fibroblasts within a loose myxoid-to-collagenous stroma. Immunohistochemically, neurofibroma is positive for S100 but negative for SMA [8]. Nodular fasciitis is a benign, self-limiting myofibroblastic proliferation composed of haphazardly arranged, plump, spindle-shaped myofibroblasts on an edematous or densely collagenized stroma. The stroma frequently harbors lymphocytes, extravasated erythrocytes, and scattered osteoclast-like giant cells [9]. Thorough examination of the current case did not reveal any inflammatory cell infiltrates, giant cells,

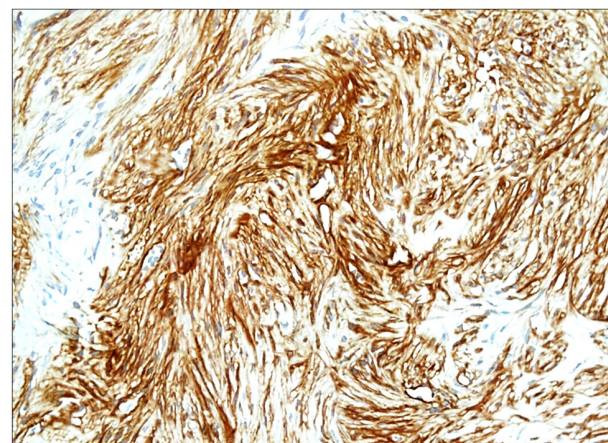


Figure 2. Diffuse and Strong Expression of SMA in the Proliferated Spindle Cells, (immunostained slide, $\times 200$).

or extravasated erythrocytes.

Hemangiopericytoma typically shows a monophasic pattern of proliferating, ovoid-to-spindle-shaped cells with indistinct cytoplasmic borders and hyperchromatic nuclei; these cells surround a rich network of prominent, thin-walled, branching blood vessels with a staghorn configuration [10]. Finally, the absence of long smooth muscle bundles and dysplastic nuclear features excluded leiomyoma [11] and fibrosarcoma [12], respectively.

In conclusion, the careful integration of clinical, radiological, and characteristic biphasic histopathological features allows for the definitive diagnosis of solitary infantile myofibromatosis, successfully distinguishing this benign condition with a highly favorable prognosis from other spindle cell mimics.

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Authors' contribution

Conception or design; Maisa hashem mohammed.

Acquisition, analysis or interpretation of data; Maisa hashem mohammed.

Drafting the manuscript; Maisa hashem mohammed.

Approval of the manuscript version to be published; Maisa hashem mohammed.

Agreement to be accountable for all aspects of the work; Maisa hashem mohammed.

Conflict of interest

The author declares that she has no conflict of interest to disclose.

Data availability

Included in the manuscript.

Ethical considerations

This case report was approved by the Ethics Committee of Sohag Faculty of Medicine, Sohag, Egypt. Verbal consent was obtained from the patient as she was discharged from the hospital one day after curettage of the mandibular lesion.

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Study registration

Not applicable.

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