

BRIEF ARTICLES

A Rapidly Enlarging Solitary Infantile Myofibroma: A Case Report and a Review of Current Monitoring Guidelines

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ABSTRACT

Infantile myofibromatosis is a rare disorder of mesenchymal cell proliferation that can affect the skin, bone, muscle, and viscera. We present a case of a 6-week-old male with a rapidly enlarging congenital solitary infantile myofibroma. The differential for congenital tumors of the head and neck is broad, and thorough evaluation is required to rule out life-threatening malignancy. Currently, there is no first-line imaging modality of choice to assess for skeletal and/or visceral involvement in patients with infantile myofibromatosis. We recommend the use of whole-body magnetic resonance imaging (MRI), as it quickly provides detailed information regarding extent of disease and does not expose the patient to the harmful effects of radiation.

INTRODUCTION

Infantile myofibromatosis (IM) is a mesenchymal disorder characterized by a fibrous proliferation that can affect the skin, bones, muscle and viscera. Once classified as fibroblastic/myofibroblastic in origin, these neoplasms have recently been reclassified as pericytic tumors.¹ There are three distinct clinical subtypes of IM: solitary, multicentric without visceral involvement, and multicentric with visceral involvement (generalized).² IM usually presents before age two, with 50% of solitary forms and 90% of multicentric forms being congenital.^{2,3} The remaining 50% of solitary forms may present in older children and adults and tend to have a better prognosis.^{2,3} The estimated incidence of IM is between 1 in 150,000 to 400,000 live births.⁴ Typically, IM presents as a firm, subcutaneous nodule on the head and neck.

Imaging is mandatory, in order to assess for multicentric and visceral disease, as both morbidity and mortality increase with more extensive involvement.

CASE PRESENTATION

A 6-week-old Caucasian male with a history of a congenital violaceous nodule on the left occipital scalp presented to the emergency department after the lesion rapidly increased in size and subsequently ulcerated. The mass drained copious amounts of foul-smelling, purulent discharge. The patient's mother reported a measured fever of 101.7 degrees Fahrenheit, vomiting, and fussiness. He was given a dose of acetaminophen and ceftriaxone at an outside emergency department prior to transfer to our facility. Physical examination revealed a 5cm x 5.5cm firm, non-fluctuant, violaceous nodule with overlying

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Figure 1. Violaceous 5cm nodule with overlying hemorrhagic crust on the left occipital scalp.



hemorrhagic crust (Figure 1). There was no occipital or posterior cervical adenopathy. The remainder of the examination was unremarkable. A complete blood count was notable for a white blood cell count of $15.9 \times 10^9/L$ and a platelet count of $673 \times 10^9/L$.

Grey scale and Doppler ultrasound were nondiagnostic. Non-contrast computed tomography (CT) revealed no evidence of erosion of the mass into the occipital bone (Figure 2a-d).

Histological examination revealed a dermal proliferation of spindled cells with elongated oval nuclei and indistinct eosinophilic cytoplasmic borders. The spindled cells were arranged in short fascicles intermixed with thin strands of collagen bundles. Nuclear pleomorphism and mitotic activity were absent. The KI-67 proliferation index was approximately 5-10%. Immunohistochemical (IHC) staining demonstrated positivity for smooth muscle

actin (SMA) and negativity for desmin and myogenin. (Figure 3a-d). These findings confirmed a diagnosis of congenital infantile myofibromatosis. Cultures returned positive for methicillin-sensitive staphylococcus aureus (MSSA).

Further screening was recommended to rule out the presence of multicentric and visceral disease. A chest x-ray, a skeletal survey and an abdominal ultrasound demonstrated no evidence of systemic involvement (images not included). The patient underwent treatment with intravenous nafcillin and was later transitioned to oral cephalexin for a total duration of 14 days. A whole-body MRI and surgical removal of the mass were recommended as an outpatient. The mass was surgically excised one month later.

DISCUSSION

Of the three clinical variants of IM, the solitary presentation occurs predominantly in males and typically involves the dermis, subcutis, or deep soft tissues.^{3,5} The multicentric presentation consists of multiple lesions with involvement limited to the skin, subcutaneous tissues, muscles, and bones.⁶ Involvement of bone has been reported in 17-77% of patients with the multicentric form, but is only seen in approximately 5% of those with solitary myofibromas.^{2,3} The generalized form is characterized by the involvement of the skin and viscera, which frequently results in organ failure and death.⁶

The differential diagnosis of congenital tumors of the head and neck is broad and includes: dermoid cyst, Langerhans cell histiocytosis, rhabdomyosarcoma, fibrosarcoma, congenital hemangioma, and cutaneous neuroblastoma. Due to the malignant nature of some of these entities, it

SKIN

Figure 2. MRI of the brain with contrast (A) T2 weighted sequence showing an isointense mass that is (B) hypointense on T1 and (C) shows peripheral enhancement; (D) SWI demonstrates foci of low intensity suggesting internal calcifications or hemorrhage.

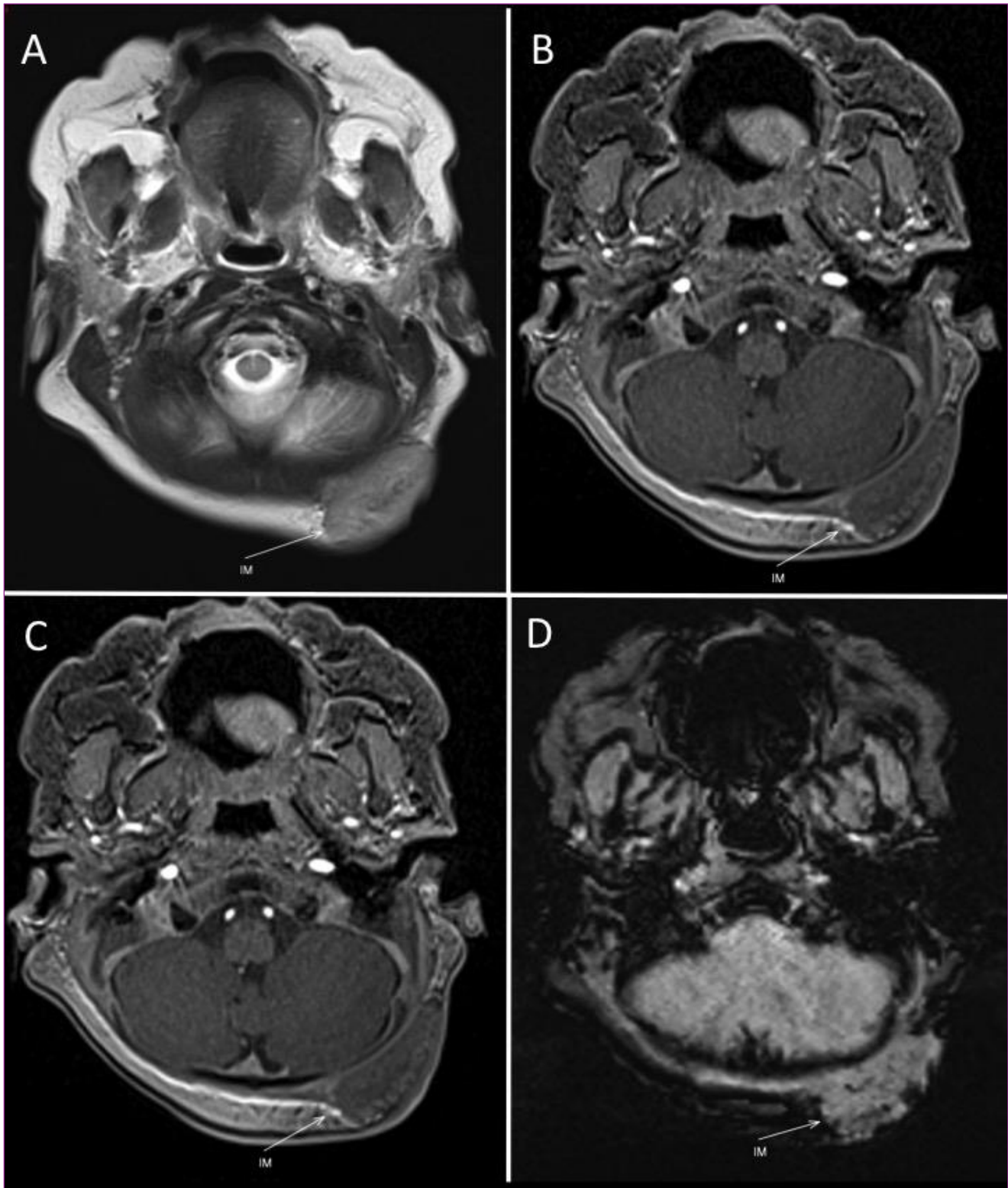
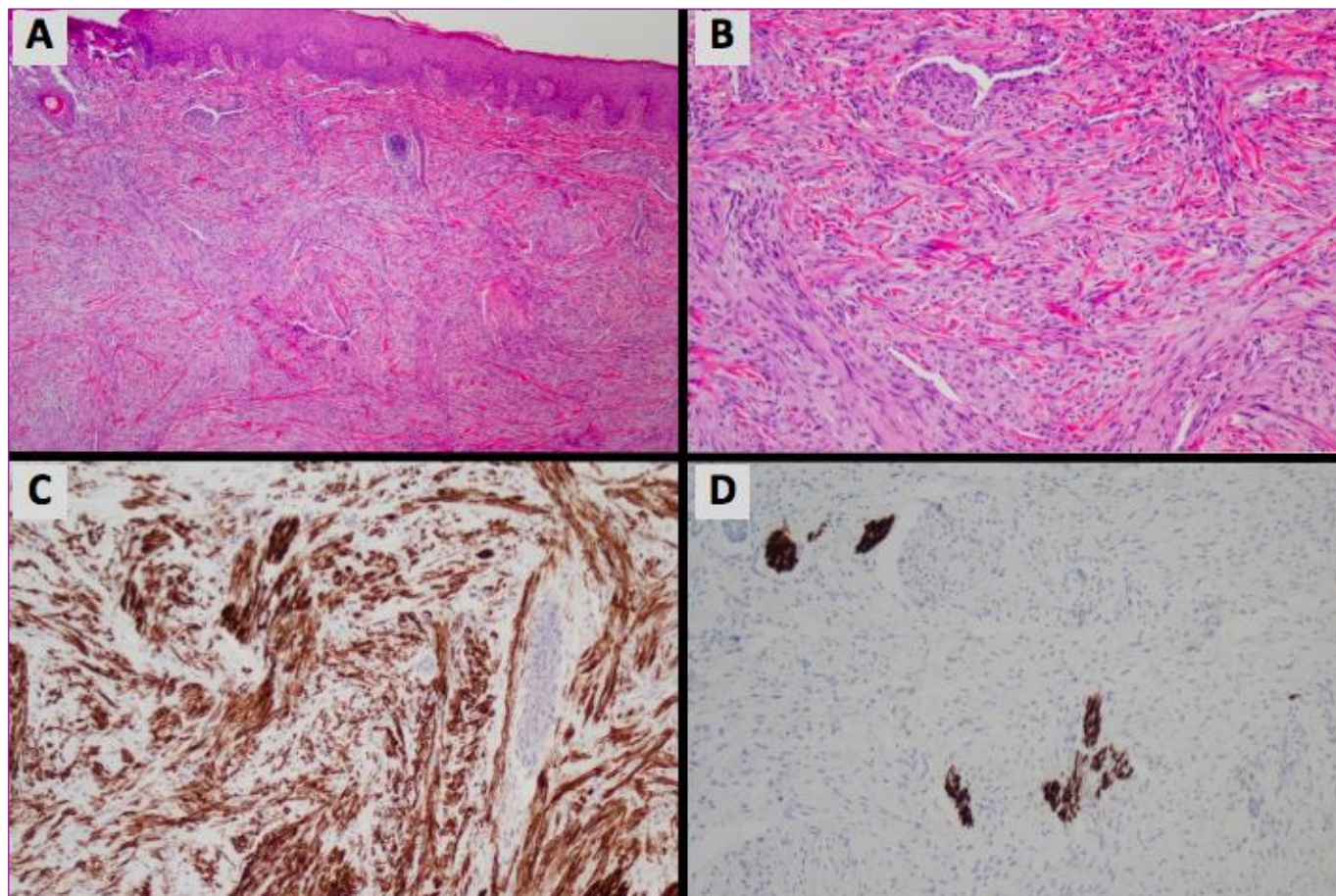


Figure 3. (A) Dermal spindled cell proliferation (H&E, 4x) (B) Short fascicles of spindled cells admixed with collagen bundles. (H&E, 20x) Immunohistochemical stains demonstrated positivity for smooth muscle actin (C) and negativity for desmin (D).



is important that all solitary nodules on the head and neck be diagnosed promptly. Definitive diagnosis can be confirmed via histopathology. Surgical biopsies of infantile myofibromatosis typically demonstrate a proliferation of myofibroblasts with pale eosinophilic cytoplasm that may be arranged in whorled or interlacing fascicles.^{2,6} Often there is a richly vascular central area that may resemble hemangiopericytomas.⁷ IHC staining is positive for vimentin and SMA and negative for S-100, epithelial membrane antigen (EMA), and cytokeratins. There is variable reactivity for desmin.⁶

Prognosis depends on the specific variant of IM. The solitary or multicentric form, in the

absence of visceral involvement, is usually associated with an excellent prognosis, with spontaneous regression occurring within one to two years.^{2,7} The generalized variant with visceral spread carries the worst prognosis. Visceral myofibromas most frequently present in the gastrointestinal tract, heart, kidneys and lungs and carry a mortality rate approaching 73%.⁴ Involvement of the heart and lungs portend a particularly poor prognosis, as these patients often succumb to cardiopulmonary failure early in life.⁷ Visceral disease may be treated with radical surgical excision in addition to chemotherapy and/or radiation.^{2,7} Treatment of solitary lesions is not typically required due to the high rate of spontaneous

regression; however, conservative surgical excision may be performed to improve a patient's quality of life if the risk for complications is minimal.^{6,9} Due to the possibility of recurrence, follow-up is essential in all patients.⁶

Once the diagnosis is confirmed, the determination of disease extent and progression with imaging is mandatory.⁷ MRI has long been considered the gold standard imaging technique for soft tissue evaluation, and is particularly adept at detecting visceral involvement in IM.^{7,10} Whole-body MRI enables a complete imaging survey within a short period of time and provides detailed information regarding the extent of disease and spread in contiguous organs.⁷ This makes it ideal to evaluate systemic involvement both initially, and every 6 months until myofibromas spontaneously regress. Additionally, MRI is an ideal imaging modality to use in the pediatric population, as it spares the patient from ionizing radiation.⁷

CONCLUSION

Infantile myofibromatosis is a rare disorder, however, its consequences can be deadly. Prompt diagnosis and subsequent imaging to assess for visceral involvement has the potential to reduce both morbidity and mortality. There are no formal guidelines regarding the imaging modality of choice for initial evaluation and surveillance. We suggest whole-body MRI as the optimal imaging modality due to its ability to assess the skin/soft tissues, bones, and viscera without exposure to radiation.

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