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Infantile myofibromatosis in a 1-month-old male from Pakistan: A case report

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Abstract

Infantile myofibromatosis is a well-recognized soft tissue tumor that is usually seen in infants and younger populations. These benign lesions mostly arise in the skin and soft tissues of the head and neck but may also involve visceral organs such as the lung, liver, and gastrointestinal tract. After a clinical diagnosis is made, a histological and immunological workup is performed to identify characteristic features like spindle-shaped cells and positive markers such as alpha-smooth muscle actin and vimentin. In most cases, since the mass is expected to spontaneously resolve, a wait-and-watch approach is adopted, especially in the pediatric age group. However, if the tumor advances to involve the organs, a complete surgical resection is carried out. In line with this, we present the case of a 1-month-old male who was diagnosed with infantile myofibromatosis after presenting with a congenital mass on the right thigh. He was born at 36 weeks of gestation to a healthy mother with an uneventful prenatal history. Imaging and histopathology confirmed the diagnosis, revealing a spindle cell neoplasm positive for alpha-smooth muscle actin and vimentin. Given the absence of visceral involvement, a conservative management approach was chosen, and the patient was scheduled for follow-up. However, the patient was lost to follow-up, which limited further assessment of disease progression. This case underscores the importance of early recognition and diagnosis of infantile myofibromatosis as a differential diagnosis for pediatric soft tissue masses. It also highlights the challenges posed by loss to follow-up in rare pediatric conditions.

Keywords

infantile myofibromatosis, pediatric, mesenchymal neoplasm, immunohistochemistry, soft tissue tumor

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Introduction

Infantile myofibromatosis (IM) is a benign tumor that mostly presents in the pediatric population. It is a combination of two words, that is, myo, implying muscle, and fibro, referring to fibroblasts that make up the connective tissue. This condition is characterized by the development of single or multiple nodular lesions in the soft tissues, bones, and/or organs, with the latter having a risk of a poorer prognosis and higher mortality. Patients typically present with a firm, rubber-like, skin-colored, or purple mass in the dermal or subcutaneous layers, but they may have an ulcerated or blood-tinged appearance.[1] The lesion measures from a few millimeters to centimeters in diameter[2] and is mostly painless. However, in case a nearby nerve becomes impinged, it can become painful and tender on examination.

Although IM is a rare diagnosis overall, with a prevalence rate of 1 in 150,000 live births, it is still one of the most common fibromas of infancy.[3] Since they can be fatal if left untreated, and no evidence-based study has reported the incidence in Pakistan yet, we concluded that reporting this case could create awareness about this disease among our population.

We present the case of a 1-month-old male who was diagnosed with IM after presenting with a congenital mass

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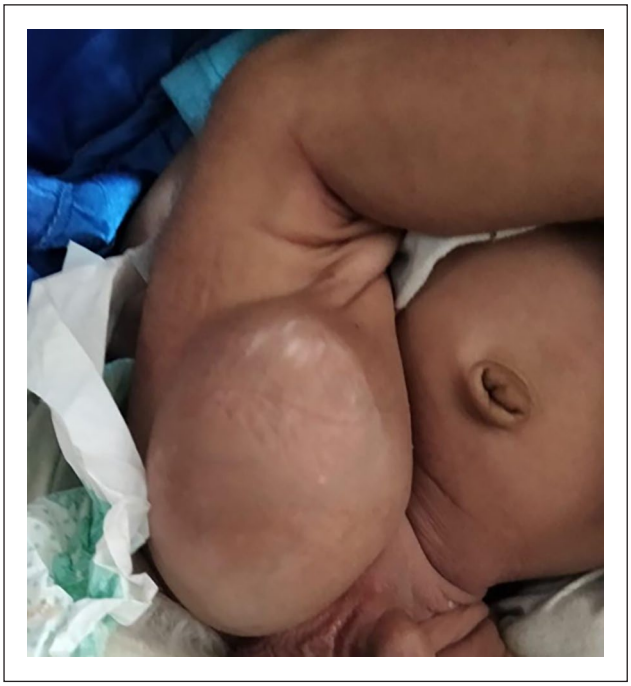

Figure 1. A solitary mass on the medial aspect of the thigh with dimensions of 4 × 4 cm.

on the right thigh in accordance with CARE (CAse REport) guidelines.[4]

Case presentation

A 1-month-old male child presented to the Pediatric Department of Dr. Ruth K. M. Pfau, Civil Hospital, Karachi, with a mass on the right thigh since birth. The child was born to a 26-year-old healthy female via normal vaginal delivery at 36 weeks of gestation. The prenatal scans were normal, and the mother did not develop any complications throughout the course of gestation. There is no history of similar complaints in the family. Moreover, parents with a history of smoking and exposure to radiation are at a greater risk of having a child with IM. The patient's parents, however, had no addictions and were nonsmokers.

Upon clinical examination, a single mass was seen on the medial aspect of the thigh with dimensions of 4 × 4 cm. The skin appeared intact with no discharge or overlying hyperemia. When palpation was performed, it was felt as a firm, sessile, non-tender swelling with well-demarcated borders. The temperature of the lesion was like the surrounding skin. The size had remained constant throughout, and there was no history of discharge or pain. There was also no associated fever, weight loss, night sweats, nausea and/or vomiting, or urinary complaints. On admission, the weight was recorded as 3.5 kg, and the child was afebrile, vitally stable, with no known comorbidity. Based on history and examination, a provisional diagnosis of IM, neurofibromatosis, lipoma, leiomyoma, and sarcoma was made. Figure 1 shows the medial aspect of the thigh where a solitary mass with dimensions of 4 × 4 cm is present.

A histopathology section of the tumor showed a small tissue fragment exhibiting a spindle cell neoplasm. The spindle cells were plumped. Focal ecstatic blood vessels were present, with nodules and fascicles of variably hyalinized cells. Immunohistochemical staining revealed the tumor to be positive for alpha-smooth muscle actin (ASMA), but negative for S100, Myogenin, MyoD, CD34, and Betactenin. Since ASMA is positive in myofibroblasts, a likely diagnosis of IM was made.

Discussion

IM is a mesenchymal neoplasm arising predominantly from fibroblasts, and bearing a biphasic growth pattern, with lightly staining bundles of spindle cells on the outside, and dark-staining areas located centrally. The rounded, immature cells in the center have plump, basophilic nuclei, an eosinophilic cytoplasm, and a delicate branching network of blood vessels around them. The peripherally located spindle cells, in comparison, have cigar-like or tapering nuclei that are either arranged haphazardly or in patterns of whorls and nodules.[5,6] However, sometimes, these two populations of cells appear in combination, and a distinct line of demarcation may not be seen between them.

The term congenital fibrosarcoma was first coined by Williams and Schrum in 1951.[7] In 1954, it was labeled as congenital generalized fibromatosis by Purdy Stout after his study on juvenile fibromas.[7,8] He believed they were multiple nodular and multicentric lesions having a benign fibroblastic process rather than a metastatic presentation and were composed of collagen-forming spindle cells. Chung and Enzinger later termed it IM after noticing its occurrence in newborns and infants and realizing its myofibroblastic features.[9] In 2002, the WHO classified the tumors into two simple categories: myofibroma to describe the solitary form and myofibromatosis for the multicentric one.[10]

Myofibroma is a condition most frequently seen in the pediatric population. A study by Chung and Enzinger in 1981 exhibited this lesion to be younger age-centric, with 60% of cases occurring immediately after birth and 88% of them happening before the age of 2.[10] Our report is also consistent with this finding, with our patient having this lesion since birth.

As per Schrodt and Callen, myofibroma has been put into three categories:

1. Solitary type, which implies a single nodule restricted to the skin, subcutaneous tissues, bone, or muscle.
2. Multicentric type, which does not include the viscera.
3. Generalized myofibromatosis, which affects the internal organs.

Among these, solitary presentation has been seen as the most common occurrence.[6] Our study is also in line with this, with the child only having a solitary lesion hitherto. While the solitary kind has been associated with an impressive prognosis with spontaneous regression or high survival rates after excision, the generalized type involving the organs has been shown to be fatal, with a mortality rate of 76%.[11]

A literature review of similar solitary cases indicates that most lesions undergo spontaneous regression, attributed to a process of massive apoptotic cell death. However, in situations where vital organ function is affected, or if there are cosmetic concerns, rapid growth, or compression effects, surgical excision is strongly advised.[12] This agrees with our case, where surgery was recommended due to family concerns regarding the size and appearance of the lesion.

In contrast, cases with visceral involvement often require a multimodal approach, including surgery, radiotherapy, and chemotherapy. Various chemotherapeutic agents such as interferon-alpha, methotrexate, vincristine, actinomycin D, and cyclophosphamide have been used with limited success.[13] Reports in the literature suggest that spontaneous regression of visceral lesions is uncommon, making early intervention critical.

In our scenario, since the family was thoroughly concerned about the size and appearance of the lesion, surgical intervention was recommended. However, due to the child's low weight, surgery was delayed, and the mother was counseled on maintaining an average weight of 5 kg before the operation. There was a loss to follow-up after this, as the patient did not return.

Our case report also highlights the need to provide thorough counseling for follow-up to patients. While it is common for delayed interventions to occur, it is crucial for clinicians to encourage return to the clinic to ensure timely treatments and avoid unnecessary complications.

Conclusion

IM is an uncommon and unfamiliar disease among the population of Pakistan. With our case report, we aim to create awareness so that it can be considered as a differential diagnosis when examining lumps in infants. Bearing in mind IM's severity when the viscera are involved, it is also crucial to keep a close look-out during the observation period of treatment so that surgical intervention can be timely done if needed.

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Ethical considerations

Our institution does not require ethical approval for reporting individual cases or case series.

Consent for publication

Written informed consent was obtained from the legally authorized representative of the patient for the publication of this case report.

Author contributions

A.A., N.A., and M.J.S.: substantial contributions to the conception or design of the work. Drafting the work or revising it critically for important intellectual content. A.M., A.Z., E.Z., and H.H.S.: drafting the work or revising it critically for important intellectual content. Final approval of the version to be published.

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