INFANTILE MYOFIBROMATOSIS- A CASE REPORT


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ABSTRACT: Infantile myofibromatosis is a rare, benign proliferative myofibroblastic tumour which occurs mostly in infants and young children. It may present as a solitary lesion most commonly involving skin, bone, muscle, subcutaneous tissue, in head, neck and trunk, with good prognosis, or a multicentric form of infantile myofibromatosis with or without visceral involvement (heart, lung, gastrointestinal tract, and kidney) with a poor prognosis. We report a case of one month old male child who presented with a subcutaneous nodule over the trunk. The mass was excised and histopathologically, immunohistochemically diagnosed as infantile myofibromatosis.

KEYWORDS: Infantile myofibromatosis, newborn, trunk.

INTRODUCTION: Infantile myofibromatosis is a rare mesenchymal neoplasm of infancy and early childhood, typically presenting as single or multicentric nodular masses of soft tissues, bones, or visceral organs. First described by Stout in 1954, various terms such as congenital multiple fibromatosis, multiple mesenchymal hamartomas, and diffuse congenital fibromatosis have been used to describe the same entity. Chung and Enzinger used the term "infantile myofibromatosis" for the first time after reviewing 61 cases. There are three different clinical entities of infantile myofibromatosis, including (1) solitary form characterized by a single nodular lesion, (2) multicentric form without apparent visceral involvement, and (3) multicentric form with visceral involvement. Patients with visceral involvement have been reported to have poor prognosis. Indolent clinical course and spontaneous tumor regression may occur in the two forms of infantile myofibromatosis without visceral involvement. Finally, in 1989, Smith et al. and Daimaru et al. coined the terms “myofibromas” and “myofibromatosis.” This nomenclature was adopted by the World Health Organization (WHO) to describe the solitary form (myofibromas) or multicentric form (myofibromatosis).1

CASE REPORT: A one month old male child presented with a swelling over the lower back on left side. The swelling was present since birth with gradual increase in size. There were no other complaints. Family history was unremarkable. Palpation revealed a subcutaneous swelling
measuring 4x4 cm, firm in consistency. Ultrasonography showed well circumscribed cystic lesion with echogenic content, suggestive of abscess. Excision biopsy was done and specimen sent for histopathology. Post operative period was nil remarkable.

GROSS – We received a globular soft tissue mass measuring 4x4x2.5cm. Cut section was solid, homogeneous grey-white.

MICROSCOPY – Multiple sections studied from the mass showed interlacing fascicles of spindle cells in a collagenised background alternating with hypocellular areas. Smooth muscle, hemangiopericytoma like areas and fibroblasts were also seen. Immunohistochemistry was performed and was positive for Vimentin. Features were suggestive of infantile myofibromatosis solitary form.

DISCUSSION: Infantile myofibromatosis is a rare mesenchymal tumour usually found in the first decade of life with 88% of cases detected before the age of 2 years and 60% at or shortly after birth. Lesions have been found in nearly all kinds of tissues, including the bone, lip, oral cavity, central nervous system, gastrointestinal tract, lungs, myocardium, liver, and biliary tree. The most common clinical manifestation is the presence of discrete nodules in the skin, muscle, or subcutaneous tissues.

Study of 199 cases of infantile myofibromatosis by Jenkins and Cawley revealed that the multicentric form (56%) was more common than the solitary form (44%). The solitary form occurred predominantly in boys (61%) primarily in soft tissue of the head, neck and trunk, so was in our case.

Superficial skin lesions may resemble hemangiomas due to prominent tumor vascularity. In multicentric form, the number of lesions varies from 2 to 100, and they may be mistaken for a metastatic lesion. Bone and soft tissue lesions may undergo rapid growth in the early weeks after birth but then regress spontaneously during the first few years of life. Although there has been no documentation describing the mechanism of the spontaneous regression of infantile myofibromatosis, Fukasawa et al. postulated that massive apoptosis was responsible.

Microscopically, both the solitary and multicentric forms have similar characteristic appearances with a distinct zoning pattern. The peripheral areas of nodular lesions are composed of spindle cells arranged in whorled or interlacing fascicles, giving a leiomyoma-like appearance. These cells demonstrate staining characteristics of both myoblasts and fibroblasts, and frequently contain a large quantity of collagen within the surrounding matrix. In the central areas, a hemangiopericytoma like pattern consisting of cells with less differentiation is usually found. A high mitotic rate greater than 3 mitotic figures per 10 high-power field and infiltration of adjacent adipose tissue and skeletal muscle are not unusual but have no adverse prognostic significance. Myofibroblasts are mesenchymal cells with both features of smooth muscle cells and fibroblasts. Immunostaining for smooth muscle actin and vimentin, with negative staining for desmin can support the myofibroblastic differentiation of infantile myofibromatosis.

The differential diagnosis for infantile myofibromatosis includes other types of fibromatosis, congenital infantile fibrosarcoma, infantile hemangiopericytoma, inflammatory myofibroblastic tumors, fibrohistiocytic tumors with a predominantly fibrous pattern, smooth muscle tumors such as leiomyomas and leiomyosarcomas, neurogenic tumors such as neurofibromas and low-grade malignant peripheral nerve sheath tumors, and nodular fasciitis.
Myofibromatosis has a number of clinical and morphologic similarities with infantile hemangiopericytoma. These authors proposed that infantile hemangiopericytoma and myofibromatosis represent different stages of maturation of a single entity, a contention supported by others.

The pathogenesis of these tumors remains unknown, but its association with estrogen receptors has been postulated. Familial occurrence has been reported in all 3 forms of infantile myofibromatosis. Most cases are sporadic, but autosomal recessive, dominant, and polygenic modes of inheritance have been postulated. Jennings et al suggested autosomal dominant inheritance with reduced penetration. Venencie et al proposed the hypothesis that infantile myofibromatosis is an autosomal recessive condition after the observation of seven siblings. These indicate a need for taking a complete family history in these cases. Cytogenetic and fluorescence in situ hybridization analyses revealed a pseudodiploid karyotype with an interstitial deletion of the long arm of chromosome 6 del (12q15q), which has been the sole anomaly found.

Whether this rare tumor is a true neoplasm or a reactive change is under investigation. Infantile myofibromatosis bears a proliferative feature and some patients even died from the tumor, which support the evidence of neoplastic characters. However, spontaneous regression was observed in some cases suggestive of a self-limiting disorder with reactive nature. Modalities of treatment other than surgery include radiation therapy, local glucocorticosteroid injections and chemotherapy, which have been used with some success when all other modalities failed. Surgery is recommended only if vital structures are affected or if tissue is needed for diagnosis. A few patients with recurrent or non-resectable tumors have been treated with some success with a combination of vincristine, actinomycin-D, and cyclophosphamide with or without radiation. In a study by Jenkins and Cawley, no deaths were reported in patients with the solitary form. In contrast, 29 patients died among 112 multicentric cases. The common causes of death were cardiopulmonary and gastrointestinal complications.

CONCLUSION: Infantile myofibromatosis is a rare mesenchymal tumour. It should be differentiated from other fibrous tissue tumours because of difference in the treatment and prognosis. It should be considered in the evaluation of newborns or young infants presenting with solitary or multiple tumours. Follow up is mandatory because recurrence has been reported in the literature.

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Fig 1 Gross globular soft tissue mass /cut section grey white
Fig 2 H&E (10x), spindle shaped cells in bundles and hemangiopericytoma pattern

Fig 3 (40x) IHC Vimentin positive