Myofibromatosis: Utility of fine needle aspiration cytology in the diagnosis of an underreported entity

ABSTRACT
Myofibromatosis (MFS) was recognized as a distinct form of childhood fibromatosis. Infantile myofibromatosis (IMF) is now identified as a solitary or multicentric tumor that predominantly occurs in neonates and infants. The adult counterpart of IMF, though of rare occurrence, is identified and is known as MFS. Morphological diagnosis of MFS is made by histopathological examination of the biopsy or surgically excised mass and confirmed on the basis of specific immunoprofile. We report a case of multicentric MFS occurring in an adolescent in whom diagnosis was suggested on the basis of fine needle aspiration cytology (FNAC) that avoided surgical excision of multiple nodules. The diagnosis was later confirmed on histopathological study and contributory immunohistochemical markers. Details of the clinical features and cytological diagnosis of the case are provided to diminish the paucity of available literature on FNAC diagnosis of the rare disease.

Key words: Diagnosis; fine needle aspiration cytology (FNAC); myofibroma (MF); myofibromatosis (MFS)

Introduction
Solitary myofibroma (MF) or multicentric myofibromatosis (MFS) represent a biphasic tumor composed of mature and immature myofibroblastic cells with hemangiopericytoma-like vessels.[1] MFS is considered rare and is underreported because the lesions may not be clinically discernible and may resolve spontaneously in most cases.[2] The occurrence of MFS in unusual age groups and sites poses a problem in clinical diagnosis. Fine needle aspiration cytology (FNAC) findings interpreted in the light of clinical context and imaging studies can point toward correct diagnosis of rare, nonmalignant entity of MFS.

Case Report
A 15-year-old female presented with multiple, bilateral neck swellings, and a swelling each on the back and the abdominal wall of five months duration. History of progressive increase in size of the swellings without pain was present. There was no significant past, personal, or family history. On examination, the patient had total seven subcutaneous, nontender, mobile swellings ranging in size from 1 cm × 1 cm to 3 cm × 3 cm in both cervical [Figure 1c, inset], right paraspinal regions and the anterior abdominal wall.

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The swellings were firm-to-hard in consistency without any change in the overlying skin.

Her hematological and biochemical investigations were normal and skeletal survey revealed no abnormality. With clinical diagnosis of lymphoreticular malignancy or multiple metastases, the patient was referred for FNAC.

Most prominent neck swellings on each side, paraspinal and anterior abdominal wall swellings were sampled by fine needle aspiration. Cytology smears from all four sites showed moderate cellularity and similar cytological features. Predominantly, spindle cells with eosinophilic cytoplasm [Figure 1a], collagenous matrix, and a few small cells with scanty cytoplasm were observed [Figure 1b]. Isolated plump spindle cells showed bipolar cytoplasmic extensions and round-to-oval nuclei with fine granular chromatin without prominent nucleoli [Figure 1a, inset]. Nuclear hyperchromasia, pleomorphism, or atypicality was not seen and mitoses were sparse. Necrosis, histiocytes, or pigment-laden macrophages were absent.

Cytological diagnosis of a benign condition favoring MFS was offered with a suggestion for histological confirmation.

Ultrasonographic examination of all the swellings confirmed their subcutaneous location and ruled out the presence of enlarged lymph nodes or other swellings.

A swelling each from the neck and the back was excised and sent for histological examination. The swellings were about 2 cm × 1 cm in size, well circumscribed, and gritty-to-cut. The sections revealed intact epidermis and the dermis showed the presence of a tumor mass that comprised of spindle cells arranged in a haphazard manner with areas of hyalinization [Figure 1c]. The central tumor area showed small cells with scanty cytoplasm arranged around thin-walled blood vessels [Figure 1d]. Areas of focal calcification were seen. No pleomorphism or nuclear atypia was noted. Immunohistochemistry revealed positivity for actin, vimentin [Figure 1d, inset] and desmin negativity.

Discussion

MFS was first described by Williams and Schrum in 1951, named congenital generalized fibromatosis by Stout in 1954, and later renamed as infantile myofibromatosis (IMF) by Chung and Enzinger in 1981.[3] It was known to represent a part of a spectrum of tumors showing perivascular myoid differentiation.[4] Presently MFS is considered to be an adult counterpart of IMF and described as a benign neoplasm of the skin and superficial soft tissue.[5]

Solitary form manifests as a single swelling in the dermis, subcutis, deep soft tissue, or muscles of the head, the neck, and the trunk region followed by extremities while multiple nodules (up to 100) may occur in the muscles, internal organs, and the skeleton, in addition to the dermis and subcutis in the multicentric form.[1] Rare occurrence of MFS in the eyelids, the pinna, and the ear canal is described as case reports.[6]

Extraskeletal lesions appear as a soft tissue masses with small foci of calcification on X-ray. On ultrasonography, soft tissue lesions have a nonspecific appearance. On computed tomography (CT), a central area of low attenuation and peripheral enhancement is seen and central calcification may be identified. Bony involvement appears as well-circumscribed, multifocal lytic lesions.[3]

MFS swellings are circumscribed, rubbery firm in consistency with white-gray cut surface and vary in size. On histopathological examination, a biphasic tumor made up of aggregates of plump spindle cells with eosinophilic cytoplasm that surround less differentiated cells with scanty cytoplasm arranged around thin-walled blood vessels is seen. There is no pleomorphism or atypia. Areas of hyalinization and calcification are present.[3] Variation in tumor morphology depending on age and site of occurrence is known that
delays its morphological identification. There is a paucity of literature on diagnosis of MFS on FNAC.

On immunohistochemistry, spindle cells of MFS show positivity for vimentin, muscle-specific actin and myoglobin that supports their myofibroblastic nature. Immunohistochemical negativity for S100 protein, cluster of differentiation (CD68), and desmin excludes tumors of neural, histiocytic, and smooth muscle origin.[8,9] On ultrastructure, myofibroma displays features of myofibroblastic differentiation.[10]

Clinicopathological manifestations of MFS include multicentricity with multifocal tumors occurring within one anatomic region. There is persistence of congenital lesions into adulthood with chances of local recurrence.[4]

Characteristic cytological features consist of variable cellularity of spindle cells enmeshed in a collagenous matrix and the presence of plump cells that represent mature myofibroblasts with round-to-oval nuclei and bipolar cytoplasmic extensions. Cellular or nuclear pleomorphism of malignancy is absent and nuclear characters are bland. The proportion of small, undifferentiated cells that represent immature myofibroblasts vary from case to case. Additional findings of calcification, giant cells, or inflammatory cells may be seen.

Conservative surgical excision is curative in MF. MFS may occur in adults at unusual locations with a potential for local recurrence. Clinical course of solitary form is benign and spontaneous regression is known to occur. Prognosis is less favorable when visceral lesions are present.[1] Our patient showed partial regression of swellings that were not excised and no evidence of local recurrence 18 months after excision.

Conclusion

MFS, although of rare occurrence, should be considered in differential diagnosis of spindle cell lesions occurring over a wide age range and at varied sites.

Preoperative suggestion of MFS on the basis of FNAC is possible and may avoid aggressive surgical intervention for diagnostic or therapeutic purpose in a particular clinical setting.

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Conflicts of interest

There are no conflicts of interest.

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