

Familial multiple lipomatosis—a rare syndrome diagnosed on FNAC

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Abstract

Familial multiple lipomatosis (FML) is a very rare benign condition with the prevalence of 0.002% in general population (1:50,000). It is inherited as an autosomal dominant disease. It is more prevalent in men. It is characterized by multiple, soft-tissue swellings on hand, forearm, thigh, and abdomen. We are presenting this case of a 24-year-old man with the clinical and cytological pictures of FML with a family history to put more emphasis on its differential diagnosis of multiple symmetric lipomatosis and the use of fine needle aspiration cytology as a definitive diagnostic tool for these soft-tissue lesions.

KEY WORDS: Familial multiple lipomatosis, FNAC

Introduction

Lipoma is the most common benign soft-tissue tumor, but familial multiple lipomatosis (FML) is very rare. Studies have reported the incidence of FML to be 0.002%.^[1] FML is a rare benign condition, inherited as an autosomal disorder, and clinically characterized by numerous, discrete, well-encapsulated, round to oval, subcutaneous, rubbery masses with variable tenderness, particularly, located on forearm, upper arm, thigh, and abdominal wall. Fine needle aspiration diagnosis is a very simple, easy available tool for the diagnosis of such multiple lipomas, which, characteristically, reveal clusters of mature adipose tissue. We, herewith, report the case of FML in a 24-year-old man who presented with multiple subcutaneous swellings on forearm, upper arm, thorax, and abdomen. Similar swellings were also noted in his father and brother.

Case Report

A 24-year-old man came to our outpatient department for the evaluation of multiple subcutaneous masses on medial aspect of forearm, upper arm, thigh, and abdomen. These swellings were present since the age of 10 years. He showed no underlying disease. The swellings were not painful or pruritic. He gave the history of a similar swelling that grew in the last 2 months because of which he was frightened and came for consultation. There was no history of trauma and medication. History of similar swellings was noted in his father and brother [Figure 1]. Both of them had not sought for any treatment.

Physical Examination

Numerous, discrete, round to oval, 2–6 cm, rubbery, skin colored, subcutaneous masses were present on both the forearms, upper arm, thigh, and abdomen. There was no visible change in the overlying skin. No lymphadenopathy of axillary, inguinal, and cervical regions was reported. Slip sign was characteristically positive. On the basis of these findings, a routine complete blood count (CBC) and fine needle aspiration cytology (FNAC) from the swellings were advised. CBC showed completely normal levels. Gross findings of FNAC revealed completely oily and scanty aspirate. About two to three needle passes were taken from all the accessible swellings. Slides were stained with Leishman and hemotoxylin and eosin stains.

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Figure 1: Gross photograph of lipoma.

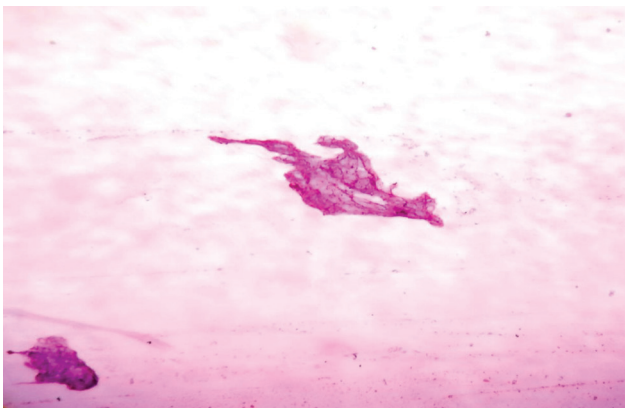


Figure 2: Microphotograph of fragments of fibroadipose tissue (H&E, 40×).

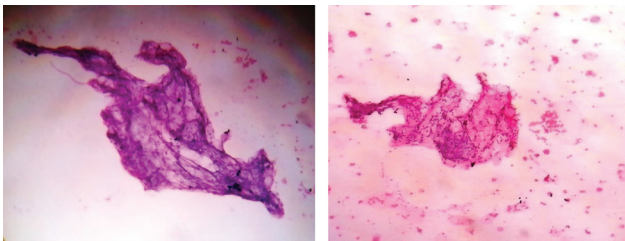


Figure 3: Both photomicrographs show mature adipose tissue with fat cells having large and empty cytoplasm with a small eccentric dark nucleus (Leishman and H&E, 100×).

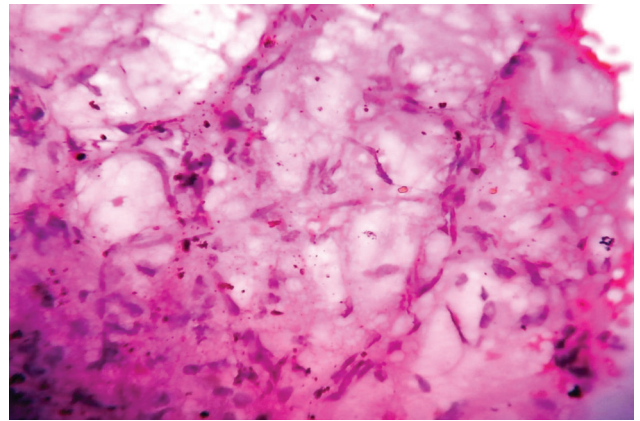


Figure 4: Microphotograph of adipocytes with abundant empty cytoplasm and a small eccentric nucleus (H&E, 400×).

Microscopic examination of cytosmears characteristically revealed clusters of mature fibroadipose tissues with a few red blood cells (RBCs) in the background. On the basis of these findings and family history, possible diagnosis of FML was given [Figures 2–4].

Discussion

FML is a very rare benign condition. It is usually transmitted by autosomal dominant group of inheritance.^[2] However, cases with recessive inheritance have also been reported.^[3] Two rare distinct familial types of lipomatosis have been identified, which are FML and multiple symmetric lipomatosis (MSL) also known as Madelung disease. Clinically, the most prominent difference between these two types of lipomatosis is that FML is marked by discrete lipomas that predominate on the extremities and generally absent from neck and shoulders, whereas MSL is a nonencapsulated, diffuse lipomatosis infiltration of underlying tissue (i.e., often is most prominent in neck and shoulder regions).^[4] The age of onset for FML and MSL are third–fifth decades and middle age, respectively. Sex predilection of FML is for male subjects, and in MSL, male to female ratio is 4:1. FML is always hereditary, usually, autosomal–dominant, whereas MSL is, usually, not hereditary. The various sites for FML are forearms, thighs, and trunk and spares shoulder and neck, while MSL affects shoulders, neck, head, and proximal upper extremity. Tumor morphology in FML is discrete, mobile with fibrous capsule. MSL is nonencapsulated and diffuse and may infiltrate deep tissue. MSL is associated with alcoholism, while FML is not. The first reported case of lipomatosis circumscript multiple, currently known as FML, was in 1846.^[5] In 1937, a case study by Murchinson describes symptoms of FML.^[6,7] Madelung was the first to describe the symptoms of MSL in 1888, by studying the disease in a man who worked in a brewery.^[8,9] In 1898, Lou Nois and Bensusade first used the term MSL to describe the vague characteristics of Madelung disease.^[10,11] In 1970, D as Gupta^[12] definitively divided benign fatty tumors into three

main categories: sporadic lipoma, FML, and MSL. Pathogenesis of FML is uncertain; some authors suggest that it is a hamartoma or mesenchymoma.^[13] FML is usually transmitted in an autosomal-dominant fashion as reported by Gologorsky et al.^[1] Male to female ratio of 2:1 shows that the male predominance is obvious. FNA is an easily accessible relatively easy to perform and less-invasive procedure, which can be used as a definitive diagnostic tool for accessible soft-tissue lesions, particularly, lipomas. Excision and biopsy can be avoided as biopsies are more invasive, painful, and costly. We gave diagnosis based on the FNA findings in our case. The main treatment of FML is the surgical excision of lesions that are responsible for cosmetic and functional impairments. In our case, no functional impairment was seen. So, the patient was advised not to undergo any surgical excision.

Conclusion

Thus, to conclude, we report the case of FML in a 24-year-old man because of its extreme rarity and its differential diagnosis to be considered from MSL and utility of FNAC as a definitive diagnostic tool for these soft-tissue lesions.

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