

Case Report



Familial Multiple Lipomatosis: Report of Two Cases

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ABSTRACT

Familial multiple lipomatosis (FML) occurring in an individual without apparent cause may be a separate disease entity or it may be a manifestation of the familial form where the family is too small to show other affected members. In this study we presented the cases of two operated brothers with FML. They were aged 61 and 47, had swelling and pain on arms, body and thigh, and difficulty in dressing. Clinical features, genetic evidence and treatment options are reviewed.

Key words: Familial Lipomatosis, Genetic transition, Excision

ÖZET

Ailesel Multipl Lipomatosis: İki Olgu Sunumu

Belirgin bir neden olmadan bir bireyde meydana gelen Ailesel Multipl Lipomatosis (AML), ayrı bir hastalık antitesi olabilir veya ailenin etkilenen diğer üyelerin görülemeyecek kadar küçük olduğu durumlarda ailesel formun bir belirtisi olabilir. Çalışmamızda AML nedeniyle opere edilen iki erkek kardeşi sunmayı amaçladık. Yaşları 61 ve 47 olan hastaların kollarda, gövdede ve uylukta şişlik, ağrı ve elbise giymekte zorluk şikayeti mevcuttu. Hastalığın klinik özellikleri, genetik bulguları ve tedavi seçenekleri tartışıldı.

Anahtar Sözcükler: Ailesel Lipomatosis, Genetik geçiş, Eksizyon

Lipomas are the most common types of soft tissue tumors with an incidence of 2.1 per 1000 people (1). FML is a rare hereditary syndrome with a proposed autosomal-dominant inheritance (2-4). Although most lipomas are sporadic, 2 rare distinct familial types of lipomatosis have been identified: familial multiple lipomatosis (FML) and multiple symmetric lipomatosis (MSL) (also known as Madelung disease) (5). In FML, lipomas are usually painless and patients are not troubled by the disease. Yet sometimes tumors grow in big sizes causing pain, difficulty in dressing and cosmetic problems (3, 6).

We investigated an affected family spanning three generations to determine the pattern of inheritance of FML. All palpable lipomas were completely excised.

CASE REPORT

Since the patients' family anamnesis revealed relatives with similar swellings, a total of 145 family members through 4 generations were contacted via telephone and 17 were found to suffer the same syndrome. The genetic pattern was examined through family tree (Figure 1). The patients stated that swellings appeared first in the 3rd decade. The physical examination of the

swellings

revealed elastic, moving, smooth, and painless masses. The lipomas ranged from size of a pea to approximately 1-6 cm in diameter and were clinically typical for lipomas (Figure 2, 3). The magnetic resonance imaging detected multiple homogeneous masses located under skin and surrounded by capsules. The most disturbing lipomas on right arm, forearm and left thigh in the first patients were excised using general anesthesia (Figure 4, 5). The pathology of obtained materials was examined.

It was found that six of the seven brothers and one of the two sisters of the patients suffered the disease; they had 67 offspring (36 male and 31 female), and only one son of both the eldest brother and youngest brother had FML. It was learned that patient's mother also had similar swellings, but not their father (died at his 99). It was detected that their mothers had 2 brothers and 2 sisters of whom one had FML. It was learned that their aunts had 6 daughters and 6 sons, of whom all had the disease; and these sons had 38 offspring (13 male and 25 female), of whom none had the disease. It was seen that out of 145 members of the family, 3 female and 14 male members have been

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affected by the syndrome.

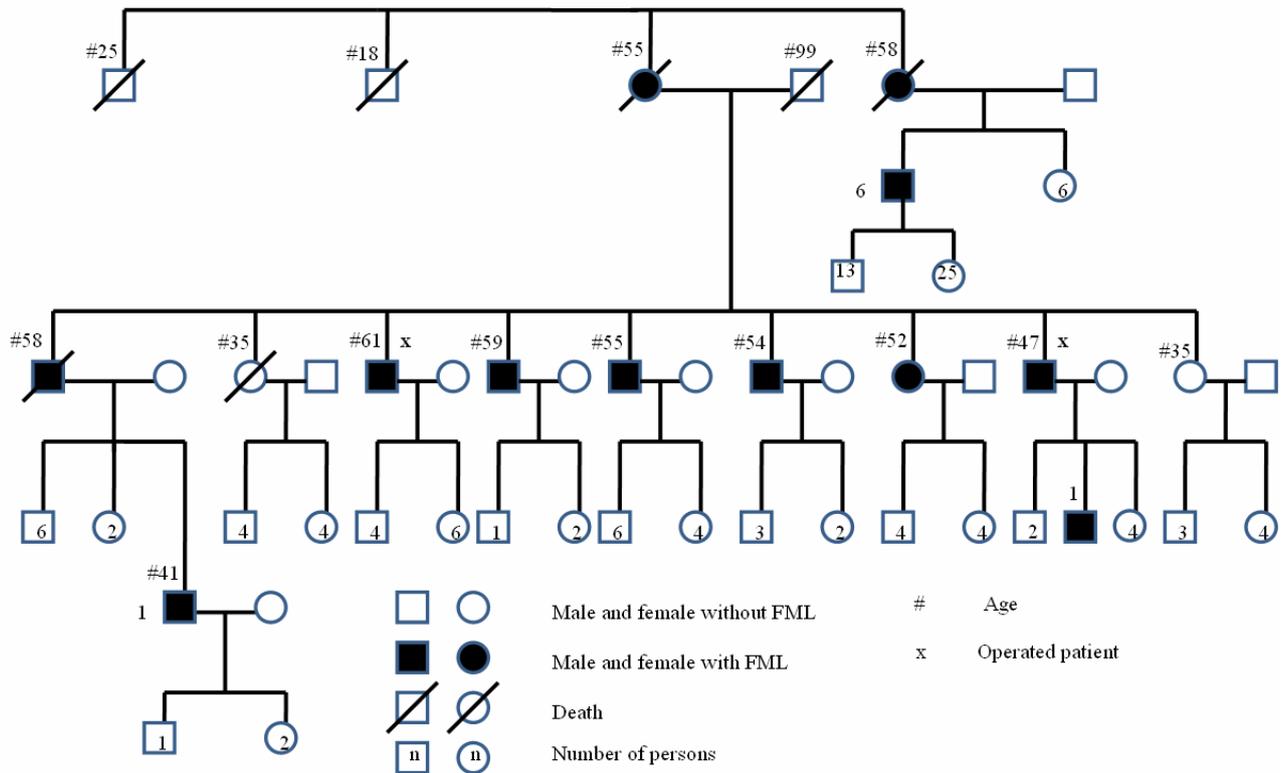


Figure 1. Pedigree of the family



Figure 2. Photograph of the operated first patient



Figure 3. Photograph of the operated second patient



Figure 4. Excised lipoma first patient



Figure 5. Excised lipoma second patient

DISCUSSION

Familial multiple lipomatosis is a very rare benign condition. It is usually transmitted by the autosomal dominant route of inheritance, although cases with recessive inheritance have also been reported (3, 7).

FML must be distinguished from MSL, which is a condition of diffuse and symmetric fatty infiltration that occurs among middle-aged men and those with a history of alcoholism (8). A distinction between the two conditions is the relative sparing of the neck and shoulders in FML and the presence of discrete lipomas rather than the diffuse and symmetric lipomatous infiltration in MSL (5, 9).

Clinical features of FML include multiple, well-encapsulated, oval-to-round, subcutaneous, rubbery lipomas that range from a few millimeters to 25

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centimeters in size (9, 10). Spontaneous regression and malignant degeneration are rare. The disease is not associated with any abnormalities in lipid metabolism (11).

Many authors state that the male/female ratio of FML is 2:1 (3, 7, 10). Some of these statements are based on previous papers and are not supported by independent data. The validity of this ratio remains unknown (4). Other authors question the validity of the 2:1 ratio and report no gender prevalence in their case studies (5, 9, 12). However, many of these reports are incomplete, as generally the disease does not manifest until after age 30. In the present study male/female ratio was found 4, 6:1.

Lipomas in FML begin to appear during the third decade of life and may continue to develop through the fifth decade of life. Lipomas are generally restricted to the arms, lower trunk, and thighs, and are asymptomatic. Patient concerns with FML usually are cosmetic. Lipomas are generally painless and do not affect the daily activities of afflicted individuals (3-6). Treatment can include simple excision, endoscopic removal, or liposuction (5). Surgical techniques have been developed to remove dozens of tumors at once and with a minimal number of incisions and scars (1).

Ersek et al (3) stated that FML is not transmitted through simple dominant or recessive genes, and the authors entertained the possibility of it being a sex-linked disease, affected by numerous ancillary variables.

FML is a rare autosomal-dominant inherited disease. Although much is known of the genetic abnormalities of sporadic lipomas, the specific germ line genetic abnormality responsible for FML is unknown. It also is not known if the same genetic abnormality is present in all families affected by FML or if different genetic abnormalities produce the same phenotype in different families. Gologorsky et al (4) did not detect any karyotypic abnormalities.

Based on the family tree prepared we concluded that the disease is autosomal dominant inherited, and also x-linked dominant inheritance is possible. Further research is needed in this respect.

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