



Macrodystrophia lipomatosa: a case report

Makrodistrofi lipomatoza: Olgu sunumu

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Makrodistrofi lipomatoza, doğuştan sınırlı jigantizmin nadir bir türüdür. Sağ ayak başparmağında şişlik, şekil bozukluğu ve ayakkabı giymede zorluk şikayeti olan 40 yaşındaki erkek hastada, konvansiyonel röntgen, ultrasonografi ve bilgisayarlı tomografi incelemeleri yapıldı. Radyolojik incelemelerde ayak başparmağında dorsale ve içe açılma, kemik yapıda hipertrofi ve dejenerasyon, medüller kemikte yoğunluk artışı, çevre yumuşak dokudaki kaslara infiltrasyon gösteren lipomatöz kitle izlendi. Rekonstrüksiyon amacıyla lipomatöz dokular kısmi olarak çıkarıldı. Örneğin patolojik incelemesinde düzenli yapıda yağ, bağ ve nöral doku elemanları izlendi. Ameliyat sonrası iki yıllık sürede hastanın klinik şikayeti olmadı ve lezyon alanında büyüme saptanmadı.

Anahtar sözcükler: Yağ dokusu/patoloji/radyografi/cerrahi; ayak/patoloji; jigantizm/doğuştan/tanı; lipomatozis/doğuştan/tanı; ayak baş parmağı/patoloji; bilgisayarlı tomografi.

Macrodystrophia lipomatosa is a rare form of congenital localized gigantism. A forty-year-old male patient presented with complaints of swelling and deformity in the right toe and difficulty in wearing shoes. Conventional radiographic examination, ultrasonography, and computed tomography showed dorsal and medial deviations in the right toe, hypertrophy and degeneration in bone structure, increased density in the medullary bone, and a lipomatous mass with infiltration to the adjacent soft tissue muscles. Partial removal of the lipomatous tissues was performed. Microscopic examination of the specimen revealed fat deposits, ligamentous, and neural tissue elements in normal structure. The patient had no complaints and there was no change in the size of the lesion within a two-year follow-up period.

Key words: Adipose tissue/pathology/radiography/surgery; foot/pathology; gigantism/congenital/diagnosis; lipomatozis/congenital/diagnosis; toes/pathology; tomography, x-ray computed.

Macrodystrophia lipomatosa (ML) is a kind of limited gigantism. It is usually encountered at birth. It is characterized by progressive growth of all mesenchymal tissues as well as asymmetrical enlargement of fibroadipose tissue of fingers or toes. This condition rarely affects the whole extremity. Macrodystrophia lipomatosa is accepted as non-congenital developmental abnormality.

Case report

40 years old male patient presented to orthopedics out-patient clinic with complaints of swelling

and distortion at right first toe as well as difficulty in wearing shoe to his right foot. He declared that this swelling is present from his birth and is progressively enlarging (Figure 1a). His past medical history was unremarkable. He was evaluated by conventional X-ray, computed tomography and ultrasonography. Conventional X-ray demonstrated soft tissue swelling at the first right toe, narrowing of the proximal and distal interphalangeal joint and degenerative hypertrophic findings similar to exostosis secondary to osteoarthritis (Figure 1b). The first right toe was showing angulation dorsally and

medially. Besides, hypertrophy was noted in sesamoid bones located in medial side of ankle. Computed tomography showed a heterogeneous lipomatous soft tissue within first right toe. The lipomatous tissue was found to infiltrate the muscular tissue (Figure 1c). Degenerative hypertrophic variations were noted in bones of proximal and distal interphalangeal joints. Though trabecular structure could be visualized in proximal and distal phalanges, differentiation of cortex and medulla was partially distorted secondary to increased density in medullar bone. Ultrasonography demonstrated echogenous lipomatous soft tissue swelling. After diagnosis of macrodystrophia lipomatosa, the first right toe was reconstructed by partial extraction of the soft tissue. Pathological examination of the material demonstrated regular adipose, connective and neural tissue elements. Patient did not have any complaints during 2 years of follow-up postoperatively.

Discussion

Macrodystrophia lipomatosa (ML) is a developmental anomaly which has also been called as partial acromegaly, macrosomy, elephantiasis, megalodactyly, dactylomegaly, macrodactyly and limited gigantism. It has been first described by Feriz in 1925 as a partial gigantism of lower extremity. Golding suggested in 1960 that the same nomenclature could be used for upper extremity lesions too. Progressive and static types of the disease have been reported.

Although the certain etiology of the disease has not been known yet, lipomatous degeneration, fetal circulation abnormality, and damage of extremity bud in intrauterine life are some of the proposed hypothesis.

Deformity is realized just after delivery. The condition may affect more than one finger; even it may involve the whole extremity. The most commonly involved parts are the second and third fingers of the upper or lower extremity and the fifth finger is very rare. Bilateral involvement is very rare. Premature closure of epiphyseal ossification centers of phalanges and metatarses, syndactyly, polydactyly, clinodactyly, brachydactyly and symphalangism may accompany ML. Involvement of lower extremity is encountered more frequently than involvement of upper extremity. There is no gender predilection in ML. Growth velocity differs from one patient to

another, even from one finger to another. The involved finger get enlarges to width and to length. Sometimes only growth in length is seen without accompanying growth in width. Multiple small osseous prominents imitating osteochondromas and

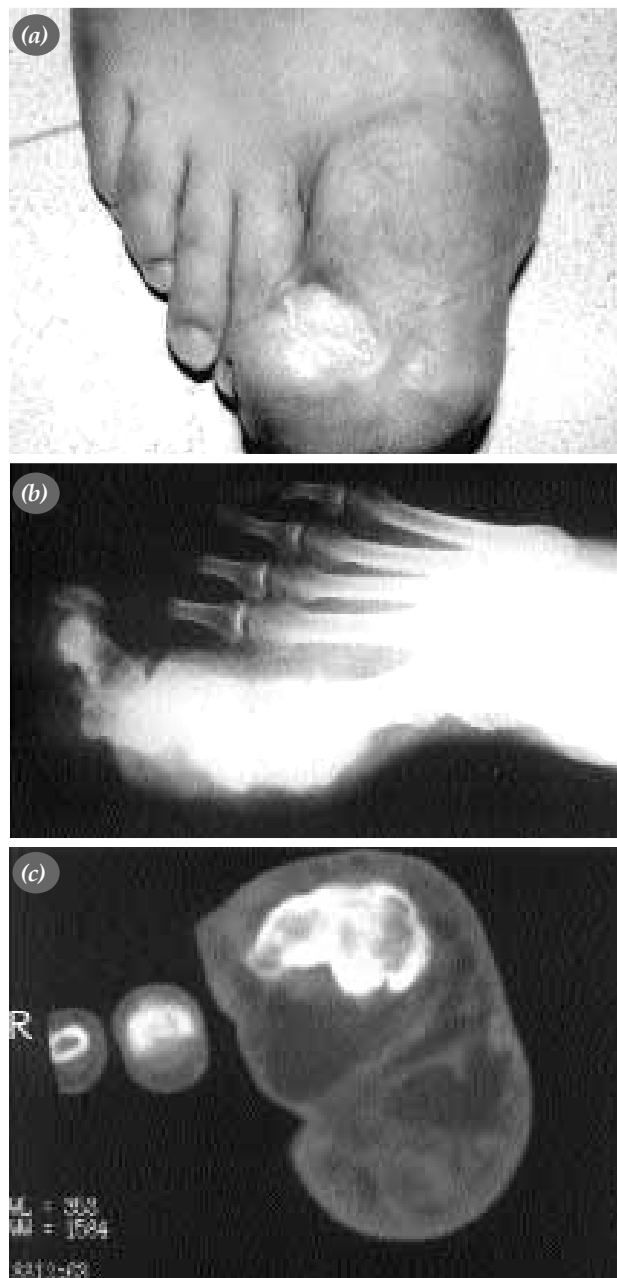


Figure 1. (a) Soft tissue swelling with medial and dorsal angulation in right first toe. (b) Soft tissue swelling and degenerative hypertrophic lesions secondary osteoarthritis similar to exostosis in phalanges seen in X ray. (c) heterogenous lipomatous soft tissue mass in fat density within right first toe visualized by CT.

osteophytes are found in the involved bone. Narrowing of joint space, subchondral cyst and large osteophytes may develop secondary to osteoarthritis.

Abnormal growth of the involved finger ceases in puberty. Surgical intervention is usually carried out due to cosmetic reasons. However mechanical problems may develop secondary to degenerative joint disease causing impairment of function or development of neurovascular compression due to large osteophytes. Giving proper information about the nature of the disease is essential before performing surgical intervention.

The most eye-catching pathologic sign is visualization of abundant adipose tissue within a net of fibrous tissue. Bone marrow, periost, muscles, neural sheath and subcutaneous tissues may get involved by ML. Neural enlargement may ensue secondary to infiltration of neural sheath by fibroadipose tissue without an increase in number of axons. Involvement of median nerve in upper extremity and plantar nerve in lower extremity is seen frequently. Carpal tunnel syndrome may develop in late stage secondary to median nerve involvement.

Typical X ray findings of ML includes excessive growth of soft tissue as well as osseous tissue, presence of radiolucent areas due to presence of adipose tissue and degenerative joint disease. Excessive growth of the bone within the area innervated by nerve and fat tissue proliferation within muscle fibers are the characteristic findings detected by CT. Excessive growth in soft tissue is seen in distal tip and volar faces of fingers. This results in dorsal angulation in the involved region. Radiolucent areas within increased soft tissue shadows are due to adipose tissue. Widening at the distal tip of bones leads to mushroom-like appearance. Decrease in diameter of bones is rare. Such growth abnormalities can also be seen in conditions like vascular steel phenomenon. In this condition, growth of fingers is caused by increased blood circulation to the finger. Gupta et al. have detected that the trabecular structure of bones are characteristically well-preserved despite growth of fingers in width and in length. MRI can easily demonstrate the fatty infiltration of the muscles as well as osseous hypertrophy and cortical thickening in the affected part of the body. Adipose tissue within the lesion is at the same density as the subcuta-

neous tissue. Fibrous striation within the adipose tissue can be visualized as low signal density. Differential diagnosis of ML includes neurofibromatosis type 1 (plexiform neurofibroma), fibrolipomatous hamartoma (FLH), lymphangiomas, hemangiomas and Klippel-Trenaunay-Weber syndrome, Mafucci syndrome, Ollier disease as well as Proteus syndrome. There is no familial predominance or neurocutaneous involvement in ML as seen in neurofibromatosis. Hyperintense mass lesion in T2 weighted MR images are seen near nerves in neurofibromatosis. Besides involvement in neurofibromatosis is usually bilateral and involvement of distal phalanx is not severe. Significant skin pathologies like capillary hemangiomas and varicose veins are seen Klippel-Trenaunay-Weber (KTW) syndrome. Although Mafucci Syndrome resembles KTW syndrome, soft tissue varicose veins are not seen in Mafucci syndrome. Diffuse swelling and pitting edema are present in cases of lymphangiomas. It is expected to see warm-like high density areas in T2 weighted MR images in cases of hemangiomas. However in both lymphangiomas and hemangiomas, osseous growth is not seen. Adipose tissue accumulation in neural sheath is expected in fibrolipomatous hamartoma however adipose tissue accumulation in ML is present throughout the involved part of the extremity. In both conditions, histopathologic changes in the peripheral nerve are not differential. Macrodactyly is sometimes seen in fibrolipomatous hamartoma. In these cases, it may be unnecessary to define the lesion as a pathologic entity different from ML. Silverman and Enzinger have detected macrodactyly in 7 of 26 cases. Dermatologic findings as well as palmar and plantar cerebriform thickening present in Proteus syndrome is not seen in ML. Besides, lipomatous accumulations may be seen in intestines and in other tissues in ML cases. In addition, calvarial abnormalities, pigmented nevus and pulmonary cysts may be seen in ML.

In conclusion, ML can be easily diagnosed by characteristic radiologic findings except for cases who have fibrolipomatous hamartoma with accompanying macrodactyly.

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