A case of macrodystrophia lipomatosa presenting with pain

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Introduction

Macrodystrophia lipomatosa (ML) is a non-inherited developmental abnormality, resulting in the overgrowth of mesenchymal elements such as bones, fat, phalanges, nerves, tendons, and vessels and usually presenting at birth[1,2]. Involvement is commonly unilateral with the lower limbs more commonly involved than the upper limbs[2]. The second and third digits of the hands and feet are frequently involved[2].

Case report

A 10 year old girl presented with a complaint of swelling of right foot and middle 3 toes since birth. Initially the swelling was small and gradually increased and attained the present size. The swelling was not painful initially, but for the last 6 months patient had pain in the right medial side of the foot while walking. Patient also had difficulties in playing and putting on her footwear which hampered day to day activities. Pain did not radiate to any joint. Birth and developmental histories were normal. There was no family history of similar complaints or of a neuro-cutaneous syndrome. On examination, swelling was present on right foot with swelling of all toes but especially on second, third and fourth toes (Figure 1)

On palpation, the toes were bulky and tender with firm skin on plantar aspect of the enlarged toes. There were no overlying skin alterations or pitting oedema and no signs suggestive of a neuro-cutaneous marker. Rest of the systemic examination was normal. Plain radiography of the right foot revealed enlarged second, third and fourth toes and hypertrophy/enlargement of phalanges of 3rd toe (Figure 2).

Figure 1: ***Anterior aspect of foot showing swelling of 3 toes***

Figure 2: ***X-ray right foot showing enlarged 2nd, 3rd, and 4th toes, the arrow showing increase in size of phalange of 3rd toe.***

Magnetic resonance imaging (MRI) was suggestive of accumulation of fat in subcutaneous tissue, fat being predominantly present in the plantar aspect of right foot suggestive of ML (Figure 3,) without involvement of joints, ligaments, tendons and nerve sheath.

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Figure 3: ***MRI of right foot showing fat accumulation in subcutaneous tissue, mostly in plantar aspect***

After confirmation of the diagnosis plastic surgical opinion was obtained and they advised surgical management which included debulking surgery and cosmetic surgery. Patient could not afford the cosmetic surgery and hence debulking surgery with amputation of middle three toes was done and fatty tissues were removed (Figure 4). Tissue sample was sent for histopathological examination. The histopathology was suggestive of the presence of mature fibro-adipose tissue without any involvement of the nerve sheath (Figure 5)

Figure 4: ***Amputation of middle three toes was done and fatty tissues were removed***

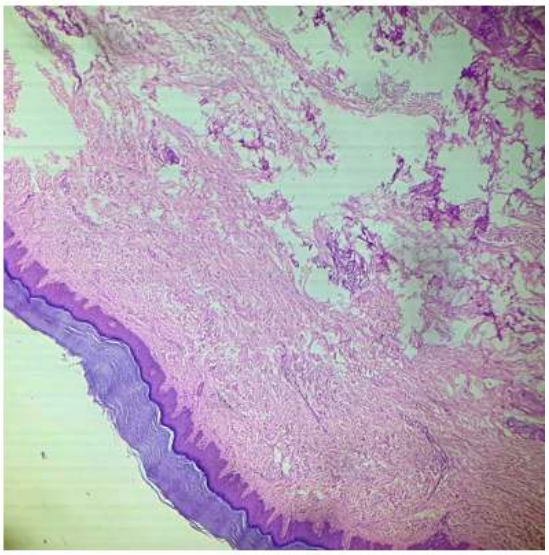

Figure 5: ***Histopathology showing mature fibro-adipose tissue without any involvement of the nerve sheath***

Discussion

There are 2 types of macrodactyly, static and progressive[3]. In the static type, the growth rate of the enlarged digits is normal whereas in the progressive type, which includes ML, the growth rate of the affected digits is increased but the abnormal growth ceases at puberty[3,4]. Aetio-pathogenesis of ML is unknown but hypotheses include lipomatous degeneration, fetal circulation anomaly, extremity bud damage, segmentation anomaly during pregnancy and increased size of affected nerve[5]. Problems are cosmetic and mechanical. In our patient pain was the most important complaint which hampered walking and daily activities. Pain was not because of neural involvement but mostly due to overgrowth of the plantar aspect, which produced dorsal deviation of the affected sole.

Diagnosis of ML can be done by various radiological evaluations such as x-ray, computed tomography (CT) and MRI. X–ray findings include excessive increase in soft tissue and osseous tissue, degenerative joint changes as reduced joint space and subchondral cysts whilst MRI reveals abundant adipose tissue and fibrous thickening of nerves[6]. As digital enlargement stops at puberty, surgical correction depends on the patient's symptoms, age and disease severity[5]. Simple debulking surgery is sufficient in most cases[7]. In our case debulking surgery and amputation of the affected toes was done.

Early diagnosis of ML either by CT or MRI and preventing mechanical complications by prompt surgical management are important. Cosmetic surgery is the best option when the swelling is small.

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