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Macrodystrophia Lipomatosa: A troubled second big toe

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We report the case of a 4-year-old boy who presented to our facility with an enlarged right second toe. After radiography ruled out acute pathology, an MRI showed increased adipose tissue at the distal aspect of the appendage. Although the parent felt that the appendage was unaesthetic, there was no functional issue, and further discussion with the pediatricians and pediatric surgeons resolved the need for immediate intervention. Macrodystrophia Lipomatosa is a rare entity that predominantly affects the pediatric population. Distributed evenly in the males and females, the disease pertains to hypertrophy of fibroadipose tissue of the distal upper and lower extremities.

Introduction

Macrodystrophia Lipomatosa is a rare entity that predominantly affects the pediatric population. Distributed evenly in the males and females, the disease pertains to hypertrophy of fibroadipose tissue of the distal upper and lower extremities.

Case Report

A 4-year-old boy presented to our facility with significant enlargement of the right second toe. The patient's mother stated that the involved digit had always been disproportionately large; however it was progressively growing in relation to the remaining digits. He had undergone resection of the distal phalanx of the second toe at the age of one year due to the hypertrophy present at that time. The working diagnosis given at that time was progressive macrodactyly of the second digit. The child did not have any other medical problems, prior hospitalizations or history of recent trauma. He maintained an adequate range of motion of the lower extremity and his second toe was not tender or warm to palpation. The mobility of the digit was slightly compromised, however he was able to move it. There did not appear to be any abnormality with his balance, although he was not entirely comfortable with his ambulation. The patient's mother complained not only of the aesthetic problem, but stated that she had to purchase two different sizes of shoes for her child. An x-ray of the affected extremity was obtained immediately to rule out an acute process (Fig. 1), followed by an MRI study (Fig. 2).

The surgery team was consulted, and after significant

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Abbreviations: CT, computed tomography; MRI, magnetic resonance imaging

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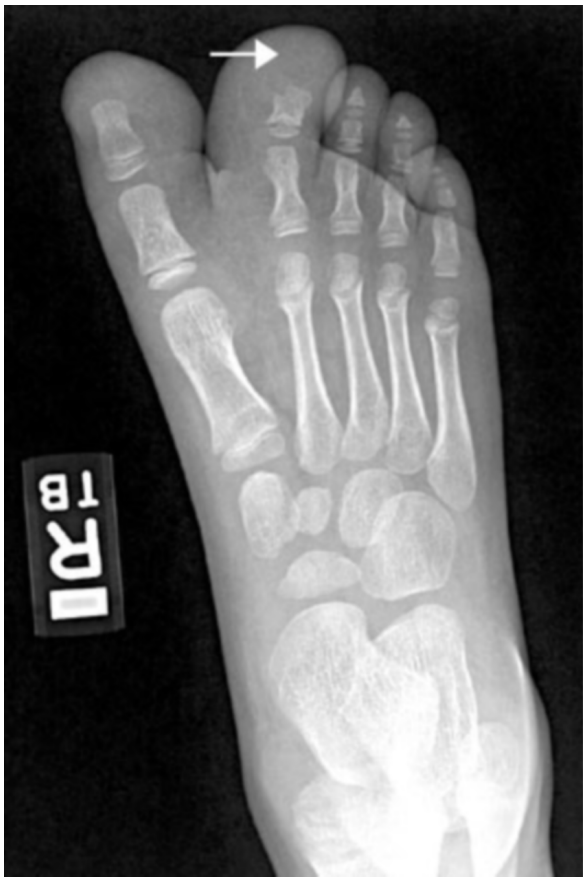

Figure 1. 4-year-old boy with macrodystrophia lipomatosa. AP radiograph of the right foot demonstrates enlargement of the soft tissues (arrow) of the second toe, which appears to be laterally deviated and approximates the first digit in size. Note is made of distal phalanx resection of the second toe with broadening and irregularity of the distal aspect of the remaining distal phalanx.

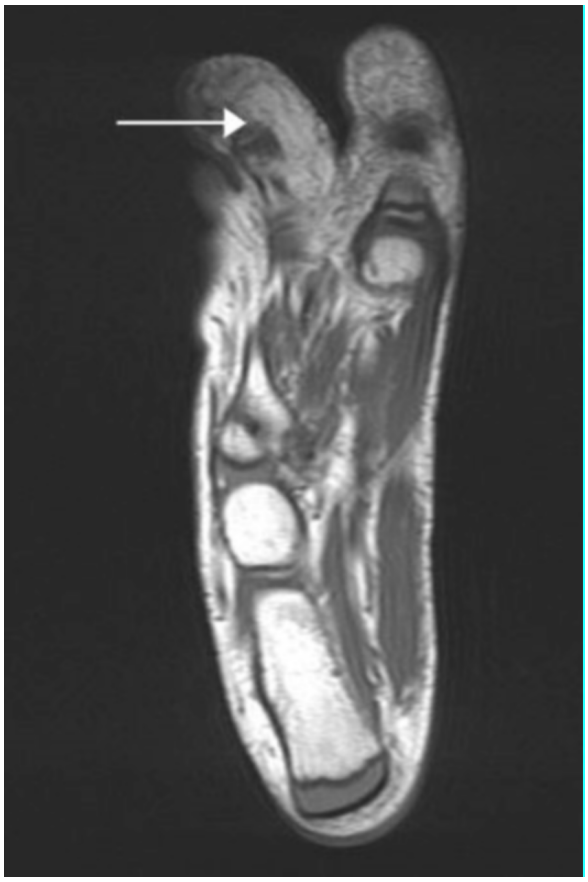

Figure 2. 4-year-old boy with macrodystrophia lipomatosa. MRI axial T1 weighted image demonstrates increased adipose tissue (arrow) around the distal aspect of the second toe, which is laterally deviated.

discussion physicians and family members determined that surgery would not be immediately offered. Pathology reports obtained from a separate institution, where the patient had undergone amputation of his distal phalanx, described an enlarged gross specimen with near normal architecture but with significant amounts of normal-appearing adipose tissue. His condition was attributed to macrodystrophia lipomatosa. Plans were made to monitor the child extensively to ensure mobility and functionality. Should these be compromised or the disfigurement become excessive, the pediatric surgeons would intervene.

Discussion

Macrodystrophia lipomatosa is a form of localized gigantism characterized primarily by proliferation of mesenchymal elements, particularly with a disproportionate increase in adipose tissue (1-3). First described by Feriz in 1925, the disease entity was expanded and has been repeatedly reported in both the upper and lower extremities (4, 5). A confirmed etiology does not exist; however, hypotheses include irregularities in the fetal circulation, lipomatous degeneration, malignant components of nerve origin altering somatic cells during embryo development, and insults to growth-inhibiting factors (6, 7, 8). However, these abnormalities were not described in our patient's histopathology, nor are they

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described in numerous other patients reported in the literature (9). Grossly, the disease affects the second or third digits of the lower or upper extremity and renders them thickened, pale, and nontender to palpation, with often impressive soft-tissue hypertrophy. Histopathological analysis revealed marked hypertrophy of adipose tissue with possible involvement of the bone marrow, periosteum, muscles, and nerve sheaths. Abnormal tissue often proliferates along the median nerve distribution of the hand and plantar region of the foot (10, 11). There have been isolated reported cases of hypertrophied adipose tissue extending beyond the extremities and into the abdomen (12). The integrity of the underlying trabicular architecture, however, remains normal (13). Interestingly, the sheer mass effect of the adipose tissue deposits can cause symptoms such as carpal tunnel syndrome, particularly when the deposits are situated beneath the flexor retinaculum and impinge on the median nerve (14).

The disease usually presents at birth and manifests as one of the two forms of true macrodactyly. The first type, coined "static," causes the involved digit to grow proportionately in relation to the other digits [15]. The second type, "progressive," involves growth of the involved digit more rapidly than the rest of the extremity, which is more in concordance with macrodystrophia lipomatosa.

Radiographic evidence of macrodystrophia lipomatosa consists of soft-tissue overgrowth prominently in the median and plantar nerve distribution. Soft-tissue radiolucency is often accompanied by elongated, thickened phalanges with splayed distal ends resembling a "mushroom" shape (15, 16). These findings are usually on the volar aspect of the digit and the respective distal end. Magnetic resonance imaging (MRI) studies have rapidly become the test of choice for elucidating the characteristic findings and helping establish the diagnosis. T1- and T2-weighted images, particularly in the coronal cross sections, reveal the proliferation of adipose tissue (17, 18). Bony or muscular abnormalities, such as cortical thickening or fibrous thickening, are also visualized using MRI studies. Sagittal short-tau inversion-recovery (STIR) images confirm the fatty lesions by signal suppression.

It is important to differentiate macrodystrophia lipomatosa from other diseases. Differential diagnoses include neurofibromatosis, lymphangiomatosis, hemangiomatosis, fibrolipomatosis of the nerve, Klippel-Trenaunay-Weber syndrome, Maffucci's syndrome, and Ollier's disease (enchondromas). Neurofibromatosis exhibits signal hyperintense neurofibromas on T2-weighted MR images, and the distal phalanges are not the most heavily affected (19). Neurofibromatosis also is associated with a familial prevalence. Lymphangiomatosis grossly presents with limb swelling accompanied with pitting edema. MR imaging reveals lesions that are hyperintense to muscle on T1-weighted images and hyperintense to fat on T2-weighted images (15). Hemagiomatosis presents with MR imaging that is hyperintense to vasculature and clinically might project an appreciable bruit. Klippel-Trenaunay-Weber syndrome consists of hemangiomas, arteriovenous fistulae, and limb hypertrophy with MRI studies revealing intermittent areas of low and high signal representing sporadic deposits of calcium and hemosiderin (8, 15). Fibrolipomatosis of the nerve is differentiated by MRI studies that reveal fatty deposits within the nerve sheath, causing marked enlargement of the nerve itself. Maffucci's syndrome can cause macrodactyly; however, it is characterized by benign enlargements of cartilage (enchondromas), bone deformities, and dark, irregularly shaped hemangiomas. Ollier's disease predominantly involves the cartilage and often encompasses various parts of the skeletal body. It is not restricted to a synchronous single extremity. Other overgrowth syndromes have been attributed to germline mutations such as Beckwith Wiedemann syndrome and Proteus syndrome. This could possibly suggest that there exists an undiscovered germline mutation leading to macrodactyly of an affected extremity (20, 21).

Cosmetic abnormalities remain a significant reason for surgical intervention in the treatment of macrodystrophia lipomatosa. Bony-growth-resembling osteophytes may lead to degenerative changes in joints, warranting operative removal for symptomatic relief. The slanting of articular surfaces leading to secondary joint disease curbs mobility by retarding joint movement and must be addressed. Furthermore, the asymmetric nature of the disease prohibits normal activity, particularly in the child population, where posture and gait are exceedingly impressionable and sensitive. Interestingly, although surgical intervention may improve immediate symptoms and fix cosmetic deformities, disease often recurs in the affected extremity. The literature has described proximal involvement of the affected extremity following distal amputation (9, 22). Liposuction has been used in attempts to "debulk" the fatty adipose

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tissue as well. Despite well-documented failures, it is imperative to address the macrodactyly, particularly among children, to prevent long-term functional disability and to keep aesthetic abnormalities from hindering social interactions and interfering with the development of healthy self-esteem.

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