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Macrodystrophia lipomatosa of the foot: A case report

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Abstract. Macrodystrophia lipomatosa is a sporadic, nonhereditary developmental anomaly and a rare form of congenital localized gigantism. It is characterized by the proliferation of all the mesenchymal elements of a digit or digits, and a disproportionate increase of fibroadipose tissue involving the nerve sheath, muscle, periosteum and bone marrow. In the present study, a 9-month-old boy was referred to Gazi University Hospital (Ankara, Turkey) with congenital unilateral enlargement of the right forefoot, particularly involving the second and third toes. X-ray and magnetic resonance imaging (MRI) scans were performed in order to assess the skeletal structures. The present study described the characteristic imaging features, with an emphasis on the MRI findings, of this rare congenital form of gigantism.

Introduction

Macrodystrophia lipomatosa is a nonhereditary developmental anomaly and a rare form of congenital localized gigantism. It is characterized by the proliferation of all the mesenchymal elements of the digits and a disproportionate increase of fibroadipose tissue. Distal limb involvement is usually observed (1-4) and the involvement of the lower limbs is more common than that of the upper limbs. Notably, the second and third digits are the most commonly affected. Macrodystrophia lipomatosa usually presents at birth or during the neonatal period (2). No gender predilection exists, however, a marginal preponderence is observed in males (1,2). Due to the rarity of macrodystrophia lipomatosa, the incidence is unknown.The etiology of macrodystrophia lipomatosa remains unclear, however, neurofibromatosis and lipomatous degeneration have been hypothesized to be involved (1). The most common treatment strategy is surgery, which primarily involves debulking and partial amputation of the affected limb (5). Patient outcome is dependent on the severity of the disease. The aim of the present study was to describe the characteristic imaging features, with an emphasis on the magnetic resonance imaging (MRI) findings, to assist the diagnosis of this rare congenital form of gigantism. Written informed consent was obtained from the patient's family.

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Case report

In March 2010, a 9-month-old boy was referred to Gazi University Hospital (Ankara, Turkey) with congenital unilateral enlargement of the right forefoot, particularly involving the second and third toes. The patient was the first child of nonconsanguineous parents, and did not present a family history of any congenital anomalies. The patient was born at term, following a normal period of pregnancy, and by an uncomplicated spontaneous vaginal delivery. At birth, the patient's weight and length were reported to be normal, and no abnormalities were noted, with the exception of right forefoot enlargement.

X-rays of the right foot reflected the macrodactyly of the second and third digits by revealing disproportionately increased length and width of the phalanxes and metatarses, and an increase in the surrounding soft tissue (Fig. 1). MRI scans identified a nonenhancing aberrant soft-tissue mass, which was hyperintense on T1- and T2-weighted images, with faint hypointense streaks on the T1 series. The mass surrounded the metatarses and phalanxes, causing the asymmetry of the foot (Fig. 2). Based on these findings, the patient was diagnosed with macrodystrophia lipomatosa and was referred for reconstructive consideration. Subsequently, the excision of excess skin and adipose tissue was performed, and the patient underwent successful bone reconstruction of the second and third digits. No complications were observed after surgery, and the patient required no further treatment. This rare case of macrodystrophia lipomatosa highlights the importance of differential diagnosis and conservative surgical treatment.

Discussion

Macrodystrophia lipomatosa is a sporadic, nonhereditary developmental anomaly and a rare form of congenital localized gigantism (1-4). This disease is characterized by the proliferation of all the mesenchymal elements of a digit or digits and a

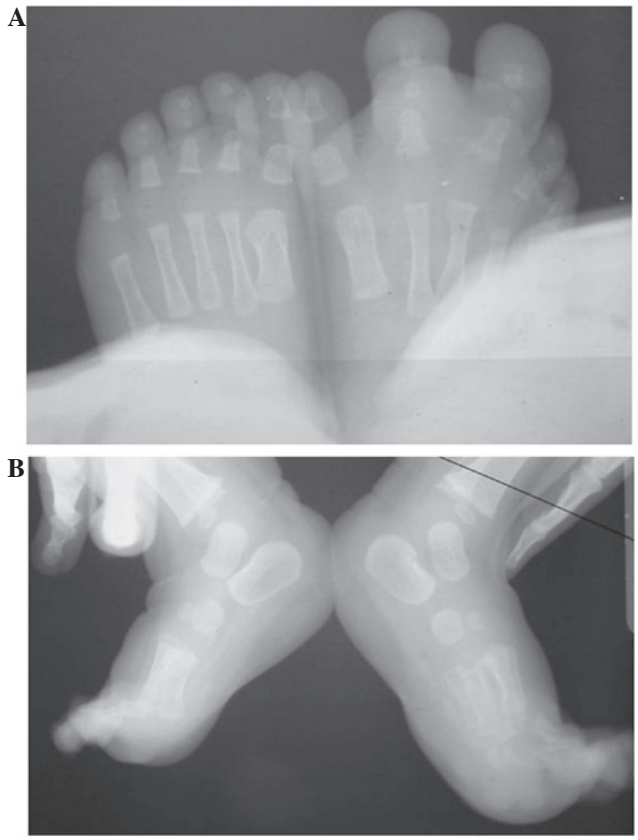

Figure 1. (A) Anterior-posterior and (B) lateral radiographic images revealing macrodactyly of the second and third digits with disproportionately increased length and width of the phalanxes and metatarses accompanied by an increase in the surrounding soft-tissue density.

disproportionate increase of fibroadipose tissue, involving the nerve sheath, muscle, periosteum and bone marrow (6).

Macrodactyly is divided into two types, including the static and progressive forms (7). In the static form, the growth rate of affected tissues is normal. By contrast, in the progressive form, which includes macrodystrophia lipomatosa, the growth rate of mesenchymal tissues is faster when compared with normal tissues and ceases its abnormal growth at puberty (4-6). Although the involvement of a single digit is common, adjacent digits are also often affected (8,9). However, bilateral involvement is rare. Secondary osteoarthritic changes, including reduced joint space, subchondral cysts and large osteophytes frequently present in adolescence or early adulthood (9). Mechanical and functional problems may develop as a result of these degenerative changes (6,10,11). An association between macrodactyly and median, plantar nerve abnormalities has also been established. Although the disease affects the nerves, it does not present with symptoms of pain (9).

Conventional radiographs have previously revealed that macrodactyly is associated with hypertrophy of the soft tissue and osseous structures. Soft-tissue overgrowth is typically observed at the distal end of the affected digits and its plantar aspect (1,2). The soft-tissue lucency detected on radiographs has been found to result from hypertrophic adipose tissue, while the phalanges are typically elongated, widened and spread out at the distal ends (3).

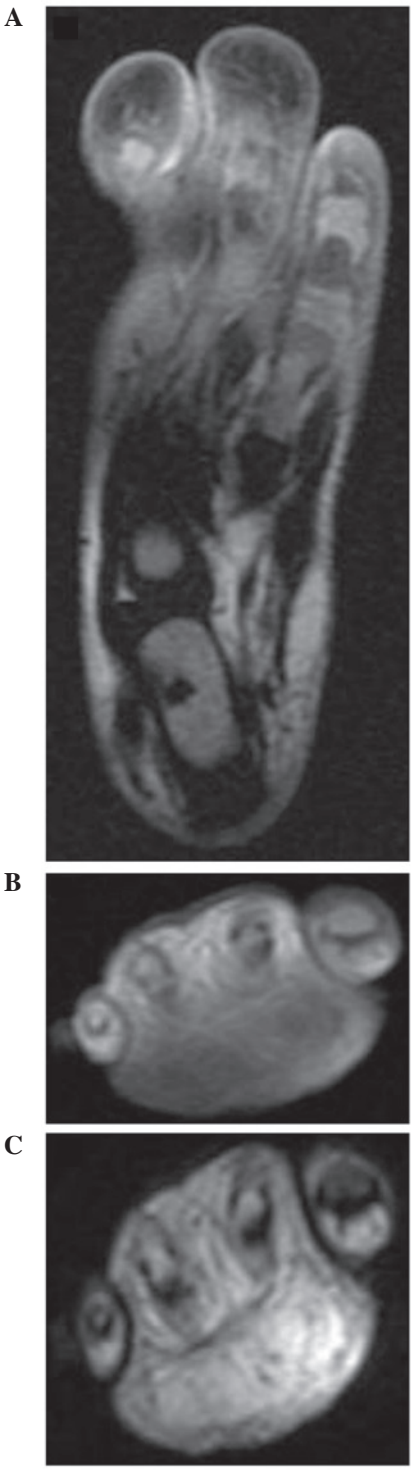

Figure 2. (A) Axial T2WI, (B) coronal T2WI and (C) coronal post-contrast T1WI revealing a nonenhancing aberrant soft-tissue mass, hyperintense on T1WI and T2WI with faint hypointense streaks on the T1 series. WI, weighted image.

MRI scans of macrodystrophia lipomatosa patients have previously revealed the presence of abundant adipose tissue, which exhibits the same signal intensity as normal subcutaneous fat in the areas affected by the disease. Furthermore, linear fibrous strands of low signal intensity within the hyperintense muscle on Tl-weighted imaging, as well as bony hypertrophy and cortical thickening, can also be detected on MRI scans of patients (4).

Based on the MRI findings, the differential diagnosis of macrodystrophia lipomatosa includes the proteus syndrome, neurofibromatosis, fibrolipomatous hamartoma, hemangiomatosis, lymphangiomatosis and the Klippel-Trénaunay-Weber syndrome (1,2,12). In the Klipel-Trenaunay-Weber syndrome, a classic triad of port-wine stains, an overgrowth of the distal digits or entire extremity involving the soft tissue, and varicose veins on the lateral aspect of the affected limb are typical signs. The disease is usually observed in children and affects a lower extremity 15 times more often compared with an upper extremity. In lymphangiomatosis, significant clinical findings include diffuse swelling and pitting edema (3). By contrast, in the proteus syndrome, hemihypertrophy is usually present, which is accompanied by lung cysts, skull abnormalities, pigmented neuvi and intraabdominal lipomas (3,8,9).

Neurofibromatosis may be challenging to distinguish from macrodystrophia lipomatosa; however, only plantar and median nerve trace affections are characteristic in macrodystrophia lipomatosa. Neurofibromatosis can present with neurofibromas, freckling in the axilla, cafe-au-lait spots and lisch nodules. Upon physical examination, hemangiomatosis may be associated with bruits, and on T2-weighted spin-echo imaging, evidence of worm-like areas of high signal intensity is common. Furthermore, fibrolipomatous hamartoma of the nerves is occasionally observed in association with macrodactyly (8).

The treatment of this disease relies on conservative surgery, which requires specific microsurgical techniques, and primarily consists of debulking and partial amputation of the affected limb. Recurrent interventions may be required for complete treatment. However, the effect of amputation on the patient's quality of life should be considered prior to surgery. Reconstructive approaches, which achieve a more aesthetic and functional result should be a preferred strategy (5).

Macrodystrophia lipomatosa is the progressive enlargement of soft and bony tissue, which leads to cosmetic and mechanical problems. The treatment of this disease depends on surgical procedures. Different imaging modalities, including radiography, ultrasonography and MRI are important for the diagnosis of this rare entity. Thus, awareness with regard to the characteristic symptoms, imaging features and differential diagnosis of the disease is important for the diagnosis and treatment of this rare entity.

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