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Macrodystrophia Lipomatosa: A Case Report

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Macrodystrophia lipomatosa is a rare congenital anomaly of localized gigantism, characterized by progressive overgrowth of all mesenchymal components, particularly fibroadipose tissue. The anomaly occurs frequently in the distribution regions of the median nerve in the upper extremity and the plantar nerve in the lower extremity. It is usually noticed at birth with predominately unilateral distal involvement of the digits. The cause of this condition is still unknown. Physicians use the patient's clinical history, physical examination, and imaging techniques to diagnose the condition, with characteristic imaging findings revealing macrodactyly with hypertrophy of the soft tissue and osseous structures. This study reports the case of a 3-year-old boy with macrodystrophia lipomatosa affecting the index and middle fingers of the left hand. Clinical presentation and radiographic imaging demonstrated typical findings, as reported in previous studies, but magnetic resonance imaging and histological examination revealed dominant fibrosis, instead of the fatty tissue characteristic to the condition. (Tw J Phys Med Rehabil 2011; 39(3): 175 - 180)

Key Words: gigantism, hypertrophy, macrodactyly, macrodystrophia lipomatosa

INTRODUCTION

Macrodystrophia lipomatosa is a rare nonhereditary local gigantism characterized by progressive overgrowth of the mesenchymal elements, especially asymmetrical enlargement of the fibroadipose tissue. It is usually recognized at birth. Tissue overgrowth often occurs along the course of the median nerve in the hand or the plantar nerve in the foot, and typically involves distal and volar regions of digits. Growth of the affected digits usually ceases at puberty.

CASE REPORT

This report describes a case of macrodystrophia lipomatosa causing progressive enlargement of the index and middle fingers of the left hand. Characteristic clinical features, along with radiographic and sonographic findings indicated the presence of the condition. However, results from magnetic resonance imaging (MRI) were dissimilar to those reported in previous investigations. This study describes this unusual case of macrodystrophia lipomatosa with atypical magnetic resonance imaging findings.
A 3-year-old boy was referred from the Department of Orthopedics for musculoskeletal ultrasound evaluation of the enlarged index finger and middle finger of the left hand. The patient was a full term baby of a healthy mother with no abnormalities during pregnancy. Disproportion among the fingers of left hand was noticed by the parents at birth, but it was not, initially, very obvious. At that time, no further assessments were made. However, disproportion among the fingers became progressively more noticeable with evidence of overgrowth of the index and middle fingers of the left hand (Figure 1). The parents did not observe any indications of pain or neurovascular symptoms and signs.

Physical examination revealed enlargement at the radial aspect of the left hand, including the thenar prominence, the index finger and the middle finger, which were non-tender and non-pulsatile. There was no pitting edema and no pigmentation was noted on the skin. As a consequence of the enlargement, the range of motion was slightly limited in terms of flexion of the index and middle fingers. The other digits' motion was not limited. The estimated muscle strength was within normal ranges in all fingers of the left hand. The patient was right-handed with normal development of hand and finger skills for a child of his age. He could use the left hand appropriately during play or activities of daily living and performed the left hand grip as expected. The lengths of the index and middle fingers of the left hand were the same as the corresponding fingers of the right hand. The largest diameter of the index finger was 5.8cm on the left hand and 4.7cm on the right one. The largest diameter of the middle finger was 5.5cm on the left hand and 4.6cm on the right one. The thumb, ring finger and little finger displayed no abnormalities. The forearm and arm were also normal. There was no family history of limb malformation or neurofibromatosis and review of the patient’s other systems detected no irregularities.

Left hand radiography was performed prior to sonography, and demonstrated enlarged proximal and middle phalanges of the second and third digits with associated prominence of the soft tissue (Figure 2). Radial deviation of the second digit and ulnar deviation of the third digit were also noted. Ultrasound revealed increased thickness of the subcutaneous tissue from the volar aspect of the left wrist, through the thenar region to the second and third digits of the left hand (Figure 3). A presumptive diagnosis of macrodystrophia lipomatosa was made according to the clinical presentation and findings from radiological and ultrasound features. MRI revealed increased soft tissues in the radial aspect of the left volar hand with hypointensity on T1-weighted images (T1WI) (Figure 4) and short tau inversion recovery (STIR) images.

Figure 1. Palmar view photographs of the affected left hand of a 3-year-old boy displaying macrodactyly of the second and third digits.

Figure 2. Posteroanterior radiographs of the left hand demonstrating prominent soft tissue and osseous overgrowth of the second and third digits.
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Figure 3. Ultrasonography of bilateral index fingers demonstrating increased thickness of subcutaneous tissue on the left side (observed on the left hand side of this figure). Left/right: 0.27/0.14cm.

Figure 4. Axial T1 weighted image demonstrating a heterogeneous nodular lesion with low signal intensity at the thenar region of the left hand.

Normal abdominal echogram eliminated the possibility of an association with Beckwith-Wiedemann syndrome with underlying tumor (for example, Wilm’s tumor). Routine laboratory tests, including hemogram and biochemistry, were all unremarkable. Biopsy of the thenar eminence and middle finger revealed fibrosis. Rehabilitation was not arranged for the patient because there were no obvious limitations to left hand function or symptoms of entrapment neuropathy. The parents were informed about the possibility of operation for cosmetic or functional reasons if abnormal growth progressed, and the mechanical problems such as entrapment neuropathy, limited range of motion, or degenerative joint disease in later life.

DISCUSSION

Macrodystrophia lipomatosa is a rare form of congenital localized gigantism with progressive overgrowth of the mesenchymal elements, especially the fibroadipose tissue.[1] The lesion often involves a single limb with predominant distal distribution. Single digit involvement is common, but adjacent digits are more often affected.[4] The affected digits enlarge disproportionally faster than non-involved digits. The anomaly is present at birth and characterized by unilateral enlarged digits.[2,5,6] Common sites of involvement are the second and third digits in both the upper and lower extremities, corresponding to the territory supplied by the median and plantar nerves.[2,7,8] The lower extremity is affected more frequently than the upper extremity. In 1992, Gupta et al reported bilateral involvement, although this is a rare phenomenon.[7] Soft tissue enlargement is caused by overgrowth of the fibroadipose tissue with variable involvement of the bone, periosteum, muscle and nerve sheath.[6] Infiltration of the nerve sheath by fibroadipose tissue can cause neural enlargement and entrapment neuropathy. There is no sexual predilection and the growth rate varies from patient to patient; even among affected digits. Digits are usually enlarged in length and width, and the terminal phalanges may demonstrate a mushroom-like appearance.[7] However, the case described in this study displayed enlargement in the width of the affected digit, but not the length.

The cause of macrodystrophia lipomatosa is still unknown. Implicated etiologies include vascular, neurological and lymphatic system abnormalities, abnormal fetal circulation, or deranged embryonal development, though there is a lack of evidence to support these theories.[9]

The characteristic radiographic findings of macrodystrophia lipomatosa are soft tissue and osseous tissue overgrowth with widened and elongated phalanges.[10,11] Marked overgrowth occurs at the distal end of the digits and along the volar aspect. MRI reveals fatty and fibrous tissue hypertrophy in the involved areas.[10] The signal intensity is intermediate to hyperintensity on T1WI, mingled with a focus of low signal intensity representing
the fibrous band, and hypointensity on T2WI, which is the same as normal subcutaneous fat. STIR images reveal signal suppression in areas of fatty tissue. MRI findings may also display fatty tissue within the nerve sheath as well as bony hypertrophy and cortical thickening. This patient’s MRI revealed marked hypointensity on T1WI and STIR images, especially at the thenar region. These findings were dissimilar to the typical MRI characteristics described in most previously reported cases. Histological examination revealed pronounced fibrous tissue with less fat, which might explain the atypical MRI features. Amounts of fat and fibrosis may vary in individual cases. Radiography revealed no obvious soft tissue lucency, which also reflected the lack of dramatic fatty overgrowth.

Diagnosis of macrodystrophia lipomatosa is usually based on clinical presentation and imaging findings including plain radiograph, CT and MRI. MRI provides a means of characterizing soft tissue for evaluation of macrodactyly and peripheral nerve tumors. Diagnostic features include macrodactyly caused by bony overgrowth, and fat deposition in subcutaneous tissue, tendons, muscles and nerves.

The differential diagnosis of localized gigantism includes neurofibromatosis type 1, lymphangiomatosis, hemangiomatosis, Klippel-Trénaunay-Weber Syndrome, Beckwith-Wiedemann syndrome, Proteus syndrome, Ollier’s disease, Maffucci syndrome and fibrolipomatous hamartoma of the nerve. The patient’s clinical history and physical examination are important factors to consider when making a diagnosis. A positive family history and ‘café-au-lait’ spots on the skin distinguish neurofibromatosis, while diffuse swelling and pitting edema identify lymphangiomatosis. The presence of small, cutaneous, red papules and nodules characterizes hemangiomatosis, along with bruits which may be auscultated. In addition, there is a hyper-intense signal on T2-weighted images in the affected area. Physicians are able to diagnose Klippel-Trénaunay-Weber Syndrome according to characteristic port-wine stains, varicose veins, and bony and soft tissue hypertrophy involving an extremity. Beckwith-Wiedemann syndrome is the most common overgrowth syndrome in infancy and the cardinal features include prenatal and postnatal overgrowth, macroglossia, and anterior abdominal wall defects. Proteus syndrome cases display characteristic partial or regional gigantism, with various other associated features such as skull anomalies, pigmented nevi, lung cysts and intraabdominal lipoma. Ollier’s disease consists of multiple enchondromas and patients present with swollen, aching limbs. Maffucci syndrome cases display characteristic benign enlargements of cartilage (enchondromas), bone deformities, and dark, irregularly shaped hemangiomas.

An occasional accompaniment to macrodystrophia lipomatosa is fibrolipomatous hamartoma of the nerve. It is a tumor-like, lipomatous lesion involving a peripheral nerve and its branches and may be indistinguishable from macrodystrophia lipomatosa when confined to the hand. Silverman and Enzinger, therefore, suggested the alternative term fibrolipomatous hamartoma of the nerve with or without macrodactyly to describe the lesion.

The most common reason for surgical intervention in macrodystrophia lipomatosa is cosmetic concern. Due to progressive growth of the lesion, multiple operations may be necessary. Debulking, epiphysiodesis and osteotomy can be considered in order to obtain the optimum outcome. The patients and their parents should be informed of a possible need for multiple surgeries. Overgrowth of digits often ceases at puberty and this should also be taken into consideration when planning surgery. Patients may experience mechanical problems later in life, such as secondary degenerative joint diseases and compression of the neurovascular structures, causing reduced joint function or entrapment neuropathy.

CONCLUSION

This study reports a case of macrodystrophia lipomatosa involving the index and middle fingers of the left hand. This case illustrates that the rare condition macrodystrophia lipomatosa should be included in the differential diagnoses of patients with localized gigantism. Diagnosis of macrodystrophia lipomatosa could be made, with some confidence, on the basis of clinical presentation and associated imaging findings, even though MRI features are atypical. It is important for the physiatrist to understand the condition’s associated musculoskeletal presentation and complaints. Familiarity with the characteristic features may also prevent further unnecessary
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testing of a patient.

REFERENCES

脂瘤性巨大症：病例報告

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脂瘤性巨大症是一種局部巨大症的罕見先天性異常，其特點是漸進性間質增生，尤其是纖維脂肪組織。該異常經常出現在上肢的正中神經和下肢的足底神經支配範圍。通常是在出生時就可發現手指或腳趾的肥大，其致病原因不明，主要是經由臨床病史，身體檢查及影像學表現特徵來診斷，顯示出先天性巨指和軟組織及骨質的增生。本文報告一位 3 歲的男孩患有脂瘤性巨大症，主要影響左手的食指和中指。其臨床表現及 X 光表現典型，但磁振造影和病理切片結果主要是以纖維化為主，脂肪組織的增生相對較少，是與其他病例報告不同之處。（台灣復健醫誌 2011；39(3)：175 - 180）

關鍵詞：巨大症(gigantism)，組織肥大(hypertrophy)，先天性巨指(macroductyly)，脂瘤性巨大症(macrodystrophia lipomatosa)