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Case Report

Fibrolipomatous hamartoma of the foot with associated macrosyndactyly: a case report ☆,☆☆

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ABSTRACT

Fibrolipomatous hamartoma is a rare benign congenital overgrowth of fibroadipose tissue in the nerve sheath. While usually affecting the median nerve, the digits of the hands and feet are sometimes affected and may result in macrodactyly, which is referred to as macrodystrophia lipomatosa. We present a rare case of fibrolipomatous hamartoma in a 6-week-old female's foot with macrodactyly and syndactyly and discuss its presentation and radiologic features.

Introduction

Fibrolipomatous hamartoma (FLH) is a benign neoplasm of the fibroadipose tissue in the nerve sheath that predominantly affects infants and children. When associated with macrodactyly of the digits in the hands or feet, this presentation is referred to as macrodystrophia lipomatosa[1].

Case Report

We present a case of a 6-week-old female infant born at full term via vaginal delivery. The patient's parents reported no birth complications, and while the patient was otherwise healthy, at birth she was noted to have significant enlargement and complete failure of segmentation of the right sec-

FLH, Fibrolipomatous hamartoma; MR, Magnetic resonance.

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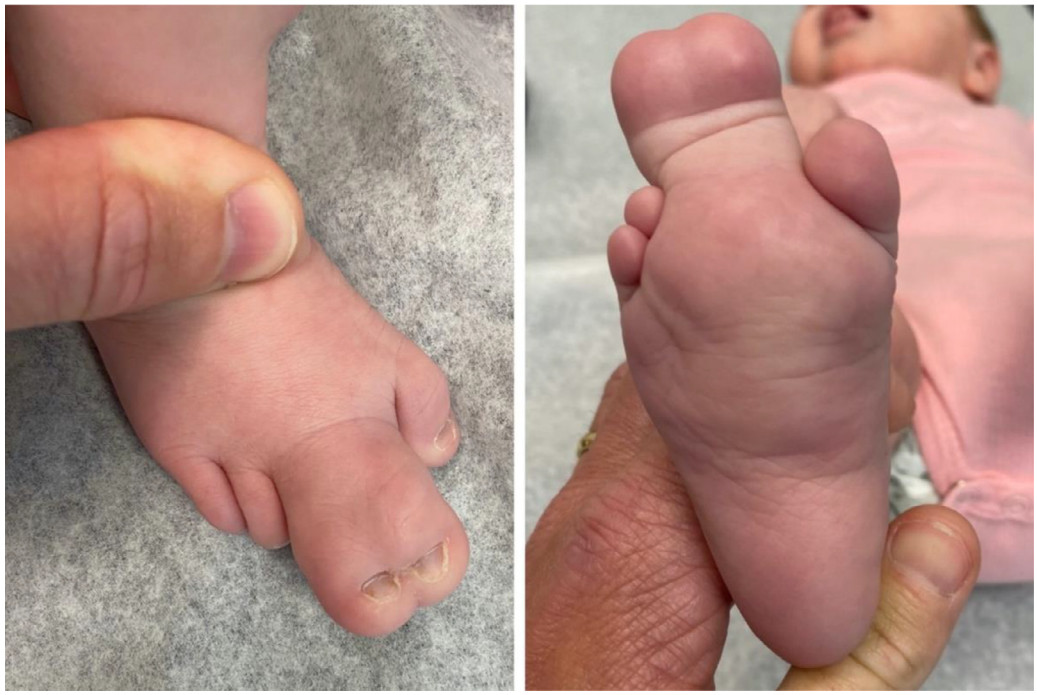

Fig. 1 – Images of the right foot of a 6-week-old female demonstrate congenital macrodactyly and complete syndactyly of the right second and third toes, and soft tissue fullness on the plantar aspect of the second and third metatarsophalangeal joints.

ond and third toes (Fig. 1). The parents reported soft tissue fullness in the plantar aspect of the foot and a tendency for the patient to hold her toes in extension. Physical examination confirmed the macrosyndactyly with note of hypertrophy of the plantar fat pad over the second and third metatarsophalangeal joints.

Plain film radiographs of the bilateral feet were obtained, which demonstrated diffuse enlargement and complete failure of segmentation of the right second and third toes with enlargement of the associated phalanges (Fig. 2A). In comparison, the left foot was radiographically normal (Fig. 2B).

Based on the clinical and radiographic findings, a diagnosis of fibrolipomatous hamartoma was made, likely involving the common digital plantar nerve in the second intermetatarsal space. The orthopedic surgeon and the patient's parents decided to continue to observe the growth and function of the digits and will consider MRI and potential future amputation.

Discussion

Fibrolipomatous hamartoma (FLH) is a rare, benign lesion characterized by fibrofatty overgrowth of the nerve sheath in extremities. First described in 1953, this lesion has been shown to involve the median nerve in about 80% of cases[2,3]. Aside from external signs, FLH may be asymptomatic in the first few years of life, presenting in childhood with painless swelling of a distal extremity or digit. This swelling progressively enlarges and may later cause pain and/or paresthesia, such as carpal tunnel syndrome in patients with median nerve involvement[3,4]. FLH may also present in the lower extremity, ulnar or radial nerves, or brachial plexus[1,2], usually in a sclerotomal distribution and unilaterally[5].

In 27%-67% of cases, presentation includes macrodactyly of one or several digits of the hand or foot, usually the second or third digit, which is referred to as macrodystrophia lipomatosa[1,4]. This rare form of localized gigantism is characterized by overgrowth of all the mesenchymal elements, including the osseous and soft tissue structures, as well as the fibroadipose tissue as seen with FLH[5]. While its etiology is not fully understood, Rios et al. have shown an association between somatic gain-of-function mutations in the PIK3CA gene and macrodactyly[6], however, it is still considered to be congenital in origin. Albeit rare, syndactyly, polydactyly, and symphalangism (synostosis of the interphalangeal joints) have been shown to be associated anomalies[5].

Imaging features are pathognomonic for fibrolipomatous hamartoma and sufficient to make the diagnosis in most cases, obviating the need for biopsy. Radiographic findings include a fat density mass in the soft tissues of the involved extremity or digits. In patients with macrodactyly, enlargement of the bones and soft tissues of the affected digits are seen[2]. Magnetic resonance (MR) imaging is the gold standard for diagnosis of FLH demonstrating an enlarged nerve with numerous hypointense tubular nerve bundles on T1-weighted images interspersed between hyperintense fat signal, commonly referred to having a "coaxial cable" appearance. Enhancement on post-contrast imaging is not usually seen[3,4]. High resolution ultrasound is a useful tool that can be done at bedside to confirm the diagnosis as well, with similar features seen as on MR; sonographic findings on transverse views show a

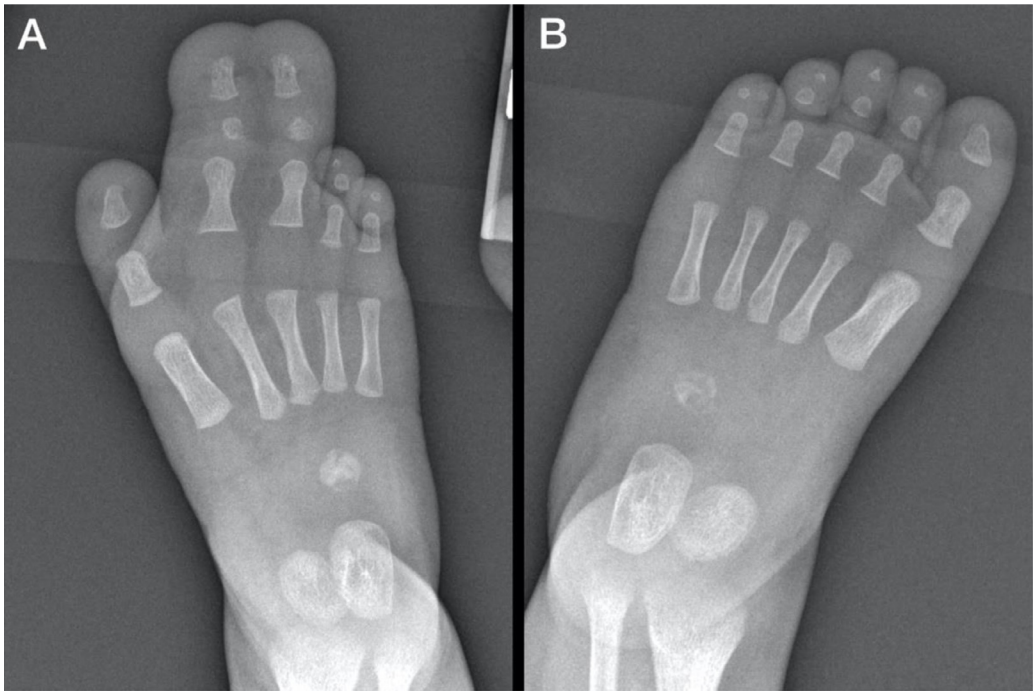

Fig. 2 – (A) Plain film radiograph of the right foot demonstrates diffuse enlargement and complete failure of segmentation of the right second and third toes with enlargement of the associated phalanges. (B) In comparison, plain film radiograph of the left foot is radiographically normal.

thickened, hyperechoic, and enlarged nerve sheath surrounding numerous hypoechoic nerve fascicles within hyperechoic fibrofatty tissue[3]. The nerves have a normal appearance and caliber in the proximal extremity, except in rare cases where the brachial plexus is involved[4].

Treatment options vary depending on the location and extent of nerve involvement, the presence of macrodactyly, and the patient's symptoms. When the median nerve is involved, conservative splinting or nerve decompression of the carpal tunnel is usually preferred[3]. Surgical options such as limited excision, ray amputation, epiphysiodesis, or soft tissue debulking are considered depending on the patient's presentation, functional limitations, and for cosmetic purposes[4,5].

Proper diagnosis of fibrolipomatous hamartoma is important as to not mistake this benign lesion with neurofibromatosis, angiomatosis, Proteus syndrome, Klippel-Trenaunay-Weber syndrome, or other rheumatological disease[3], for which additional testing or treatment may be completed unnecessarily, especially given that its imaging findings are pathognomonic.

Authorship

All authors had access and equal role in writing the manuscript.

Patient Consent Statement

Appropriate patient consent has been obtained for this case study.

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