

CASE REPORT

Surgical treatment in a case of giant macrodystrophia lipomatosa of the forefoot

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Abstract

Macrodystrophia lipomatosa is a rare, congenital, non-hereditary disease, characterized by local gigantism of the fingers or toes. We report the case of a 37-month-old boy, with no prior past medical history, who presented with a gigantic dystrophy of the left forefoot. The location of the deformity was involving the plantar and dorsal aspect of the foot, and digits 1 to 4. After clinical examination, imaging study assessment, and differential diagnosis considerations, it was established that macrodystrophia lipomatosa was the cause of the deformity. A reconstructive surgical intervention was planned. The hypertrophied tissues were excised, resulting in a reduction in the forefoot's volume. The patient had a favorable postoperative course and ambulation was allowed with a custom-made shoe. The case represents a rare pathological entity with complex diagnostic and therapeutic considerations. As far as treatment options, the surgeon must decide between a reconstructive surgical intervention and amputation.

Keywords: macrodystrophia lipomatosa, megalodactyly, debulking, local gigantism.

Introduction

Macrodystrophia lipomatosa (also named partial acromegaly, megalodactyly or limited gigantism) is a pathological condition first described, in the lower limb, by Feris in the early 1920'. This is a congenital, non-hereditary disease characterized by local gigantism [1]. Golding extended the term, in 1960, to include involvement of the upper limb [2]. From histopathological point of view, there is hypertrophy of the adipose tissue, compromising the integrity of all tissue types (like muscles, nerves, periosteum) [3–5]. Often, there is a degeneration of the bone structure and an increase in the density of bone marrow. There is also a possibility that the degeneration take over the normal structure of important nerve trunks like median and plantar nerves [5]. Treatment can vary from partial limb amputation to multiple reconstructive surgical interventions, according to the extension of the pathology (localized or extended) and the preference of the surgeon [1, 4, 5]. Taking into account the young ages of the patients, amputation should be only considered as an ultimate treatment option.

Macrodystrophia lipomatosa is a rare disease. There are less than a 100 cases reported in the literature. Most of these are relatively mild forms, involving one or two finger or toes [2, 5–9].

The low number of published cases, the lack of clear guidelines for adequate therapy and the peculiarities of the gigantic presentation of this disease justify the report of this case.

Case presentation

A 37-month-old male patient was brought by his parents in our Department, with a congenital gigantic hypertrophy of his left forefoot (Figure 1). The child had a marked impairment of his gait due to the excessive volume of his foot. No custom made footwear could be manufactured for the deformed limb and the foot was protected only by a hand woven wool sock. All previous consulted surgeons proposed amputation of the entire foot, but the parents did not accept the procedure. The deformity affected the first four toes as well as the plantar and dorsal aspect of the forefoot. The calcaneal region, ankle and calf were in normal range of development. The patient had no trauma or pathological family history. The local examination did not reveal any nervous or vascular abnormalities. According to the recommendations in the literature, the imagistic investigations included X-ray, magnetic resonance imaging (MRI) (Figure 2) and computed tomography (CT), in order to plan the operation and to exclude other conditions such as type 1 neuro-

fibromatosis, lymphangiomatosis, hemangiomatosis, Maffucci syndrome, Ollier disease and Proteus syndrome. The surgical alternatives, ranging from amputation to reconstructive techniques have been discussed with the family. After obtaining the informed consent from the parents, a reconstructive procedure, in order to reduce the volume of the forefoot, was decided. They were informed that macrodystrophy is a progressive disease, with a local postoperative recurrence rate of 33–60% until completion of growth and that for a definitive result several surgical procedures could be necessary.



Figure 1 – Preoperative aspect of the left foot.



Figure 2 – Radiological aspect. MRI demonstrating proliferation of the non-encapsulated subcutaneous fat with diffuse bony enlargement as well as widening of phalanges. Presetting evident fat deposits both plantarly and dorsally. The radiography of the forefoot reveals hypertrophy of the distal phalanges with a typical deformation mushroom-like.

The surgical procedure (Figure 3), performed under tourniquet, consisted in extensive debulking of the plantar and dorsal fat deposits of the foot. Also, the gigantic phalanges of the first four toes were resected, along with a second and third transmetatarsal amputation. The sectioned flexor and extensor tendons were sutured to each other in order to balance the corresponding muscles. With a heavy suture (FiberWire®, Arthrex, Naples, FL, USA), a connection between the first and fifth metatarsal is placed, in order to reduce the width of the forefoot

and in an attempt to restore the anterior arch of the foot. Careful hemostasis was performed. The amount of resected tissues (Figure 4) weighted about 145 g, but, in order to preserve the skin vascularization, not the entire hypertrophied fat tissue could be eliminated. In 30–50% of the cases, following overzealous debulking, local vascular complications can occur. At the end of surgery, a radical volume decrease of the foot was obtained but some excess fat tissue remained on the plantar side of the foot (Figure 5). This surplus could have been removed, if planned, by liposuction in the same session. The skin flaps were approximated with separate sutures, paying attention to the possible impingement zones between the scar and the footwear. In his postoperative evolution, the patient presented a 1.5×2 cm wound dehiscence, which healed, with local dressings, in 45 days. All the resected tissue was sent for pathological examination. The selected tissue fragments for pathological evaluation were fixed in 10% formalin and routinely processed by paraffin embedding. The sections from the paraffin blocks were Hematoxylin–Eosin (HE) stained.

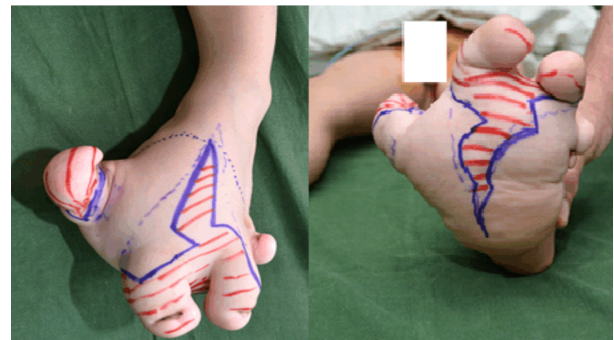


Figure 3 – Preoperative planning. The levels of the excision that allows the resection of the largest amount of degenerative tissue and to permit an adequate shape postoperatively.

The examination showed an abundant presence of fibrofatty tissue with an intense vascularization and proliferation of small nerves (Figure 6, A and B; Figure 7, A and B). After the surgery, the patient was allowed for partial weight bearing on the affected limb with progression to full weight bearing after complete wound healing. A custom-made shoe was manufactured to fit the debulked foot.



Figure 4 – Excised tissue. The amount of tissue resected that weighted around 145 g and included the phalanges of the first four fingers, large amounts of skin and parts of the second and third metatarsals.



Figure 5 – Immediate postoperative aspect. The final aspect of the sutures and the drain.

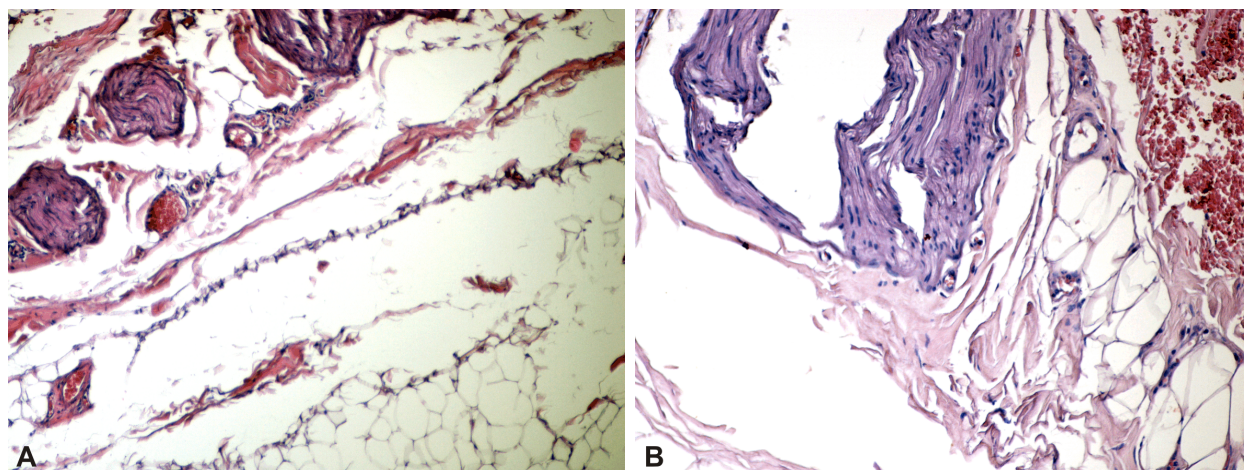


Figure 6 – (A) Fibroadipose tissue with nerves; (B) Details from the previous picture with fibroadipose tissue containing a nerve. HE staining: $\times 100$ (A); $\times 200$ (B).

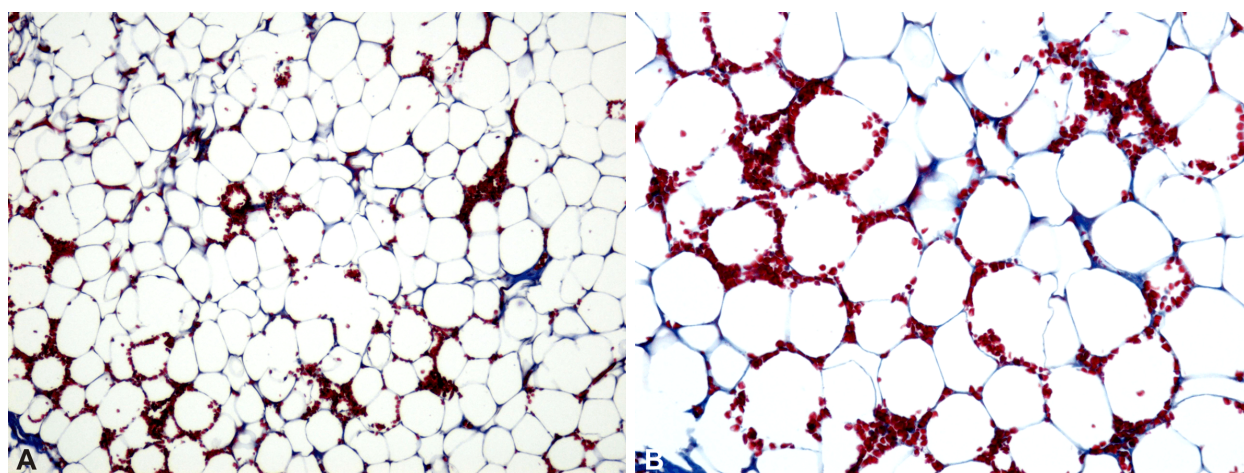


Figure 7 – (A) Area of the fatty tissue with intense vascularization; (B) Details of microvascularization of fat tissue. Goldner–Szekely staining: $\times 100$ (A); $\times 200$ (B).

Ninety days after surgery (Figure 8), the scars are healed, and the gait is almost normal. The main problem is the leg length discrepancy of 2 cm due to the still thick plantar fatty layer. This problem is now planned to be solved by liposuction as soon as the parents will agree with the treatment. Until this procedure is performed, the child will wear a pair of custom-made shoes (Figure 9) to equal leg length. It is expected that following growth, successive adapted footwear will be necessary until the final form of the foot is obtained. At the two years follow-up, the clinical status is maintained. The foot is functional and no other deformity developed. The parents are satisfied with the treatment outcome and they have not approved yet the liposuction of the plantar fatty layer.



Figure 8 – Clinical aspect at three months.



Figure 9 – The custom-made shoes.

Discussion

Macrodystrophia lipomatosa of the foot is characterized by an abnormal hypertrophy of the mesenchymal elements and can affect a toe, several toes or the entire foot [5, 8, 10]. There are a number of theories regarding the etiology of this disease (abnormalities of the fetal circulation, errors of the intrauterine segmentation, hypertrophy of the involved nerves) [11–14], but in this case no link to the mentioned conditions could be established. Although in the literature there are some differences of the reported histological features, related with the different denominations of the described lesion and with the common confusion of macrodystrophia lipomatosa and fibrolipoma of nerve, the most authors believe that the gross changes have as histopathological substrate the proliferation of mesenchymal elements with an excess of adipose/fibro-adipose tissue [6] in the dermis, subcutaneous tissue, nerve sheaths, bone marrow, periosteum and muscles [3, 15, 16]. The same aspects were also confirmed by our observations. There were described areas of hyalinosis and basophilic degeneration of collagen, proliferations of subcutaneous nerves [7] moderate atrophy of the osteo-cartilaginous tissue [17] or hypertrophy, exostosis, ankylosis of interphalangeal joints and fatty invasion of

the medullary cavity [18]. If the clinical and imaging differential diagnosis of macrodystrophia lipomatosa include numerous entities such as hemangioma, lymphangioma, Ollier's disease, Klippel–Trenaunay–Weber syndrome and plexiform neurofibromatosis (NF) [16], the morphopathological one imply the discrimination of the lesion from the fibrolipomatous hamartoma of nerve which can produce digital overgrowth and which is an isolated nerve lesion [19], a tumor-like condition that consists of an infiltration by connective tissue and fatty elements of the nerve [16]. Besides, macrodystrophia lipomatosa could be associated with fibrolipomatous hamartoma in as much as 30–66% of cases [20]. The major drawbacks of macrodystrophia lipomatosa are functional but also cosmetic [21]. Due to excessive limb volume and functional degenerations, as the patient gets older, osteoarthritic changes can evolve, resulting in nervous and vascular compression, ending up in multifactorial dysfunction [7]. Therefore, although no specific timing for the treatment is currently reported, we consider that as soon as the child can tolerate major reconstructive surgery, the first corrections of the limb should be performed. This will help with proper development of motion and corporal awareness. The surgical procedure has the main purpose to create a cosmetic acceptable result, but it also needs to restore the function of the limb [7]. The chosen technique was designed to fulfill as much as possible these requirements that also meet the expectations of the parents. The preoperative planning has considered the eventual neurovascular abnormalities, the amount of skin, fatty tissue and bone that will be resected. The skin flaps and the extent of debulking were performed according to the local vascularization anatomy, and in order to prevent postoperative wound complications that can occur in 30–50% of the cases [5]. Not all operative steps could be performed as planned. Part of the surgical technique has been decided during the intervention, as for example, the placement of the suture connection between the first and fifth metatarsal to reduce the width of the forefoot, but also, to rebuild an anterior arch to the foot. On the other hand, liposuction could have been useful during the initial intervention, but not considering it in advance made it a second intent option. As reported, the deformity is more frequent unilaterally, in the plantar nerve distribution area [20]. Usually, the lower leg is involved, and the disease is progressive, this being the reason for maintaining the fifth toe, as reference of the surrounding tissues to a normal developed segment [18]. This leads, as already stated, to a reported local recurrence rate of 33–60%. That is why, probably, several successive procedures will be necessary in order to achieve the desired outcome at the completion of skeletal growth [22, 23]. The amount and extension of the future interventions will be tailored depending on the evolution of the deformity and the degree of functional impairment. Before any surgery can be planned, skin flaps have to properly heal first. The residual differences between the two limbs should be corrected first by orthotic shoes. These will need to be custom changed following the evolution of growth and that of the possible local recurrence of the deformity and its treatment.

Conclusions

Macrodystrophia lipomatosa represents a complex, rare pathology, which needs an elaborate differential diagnosis and a full imagistic exam. The deformity is treated most often by surgical reconstructive techniques; amputation should remain as a final salvage intervention. These procedures are intended to improve the functional outcome of the affected limb, to reduce its volume and to provide an acceptable aesthetic appearance in order to ensure the social integration of the patient.

Conflict of interests

The authors declare that they have no conflict of interests.

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Received: September 3, 2016

Accepted: November 18, 2017