Progressive macrodactyly. A case report

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Case Report

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Background: Macrodactyly is a rare congenital abnormality, characterized by overgrowth of the bones and soft tissues of the fingers. The etiology is unknown although several possible mechanisms have been proposed such as abnormal innervation leading to free growth, increased blood supply and humoral mechanism that stimulates growth. The objective of surgical treatment is to obtain a non-painful and cosmetically functional foot that adapts to all terrain where the child moves. The procedures suggested in the literature are: finger amputation, epiphysiodesis and soft tissue reduction accompanied by a partial or total resection of the distal phalanx and ray amputation ("princess foot").

KEYWORDS: Macrodactyly.

Macrodactyly known as digital gigantism, megalodactyly, lipomatous macrodystrophy and fibrolipomatosis with macrodactyly is a nosological entity defined as a rare congenital deformity without hereditary transmission with the presence of a disproportionately large finger that is discovered at birth or in the first years of life given by a hyperplasia of all the elements of the affected finger[1].

It has a prevalence of 0.08 per 10,000 newborns, this is even lower when it occurs in isolation[2].

The etiology is unknown although several possible mechanisms have been proposed, such as abnormal innervation that leads to free growth, increased blood supply, and humoral mechanism that stimulates growth[1].

The peripheral nerve enlarges in both circumference and length. A new mutation has been described in PIK3CA (R115P) present in the affected nervous tissue, but not in the blood DNA. In addition, it has been possible to identify that the perineurium is the most abnormal structure within the nerve involved in macrodactyly of a finger, with additional side effects on axon number and size[3].

Mainly two types have been reported: static and progressive. Static macrodactyly is present from birth, it is sporadic and isolated, the growth rate corresponds to that of the patient and has proportionate growth. The second type is represented by progressive macrodactyly or lipomatous macrodystrophy, which is congenital and has no defined pattern of inheritance. In it there is accelerated and disproportionate growth, beginning in the postnatal period and ending with growth coinciding with the end of bone maturation. The characteristic is a growth of mesenchymal tissue and fatty infiltration of soft tissues. The predominant areas are those innervated by the median and plantar nerves. Receptor tyrosine kinase (phosphoinositol 3-kinase) and tyrosine/serine kinase dysfunction has been observed, as in other overgrowth syndromes[4].

Upton described four types of macrodactyly: type I presents lipofibromatosis of a nerve, either of a static or progressive subtype; type II is associated with neurofibromatosis; type III has associated hyperostosis; and type IV is associated with hemihypertrophy. It can also be associated with Beckwith-Wiedemann and Proteus syndromes, which although not true macrodactyly, arteriovenous malformations can present with enlargement of elements of the hands and feet, and can be treated similarly to macrodactyly[3].

The topographic location is greatest on the fingers and its second location is on the toes[2].

The objective of surgical treatment is to obtain a non-painful and cosmetically functional foot that adapts to all terrain where the child moves. The procedures suggested in the literature are: finger amputation, epiphysiodesis and soft tissue reduction accompanied by a partial or total resection of the distal phalanx and ray amputation ("princess foot"). However, in procedures involving partial amputations, the results are not satisfactory and multiple surgeries are required[2].

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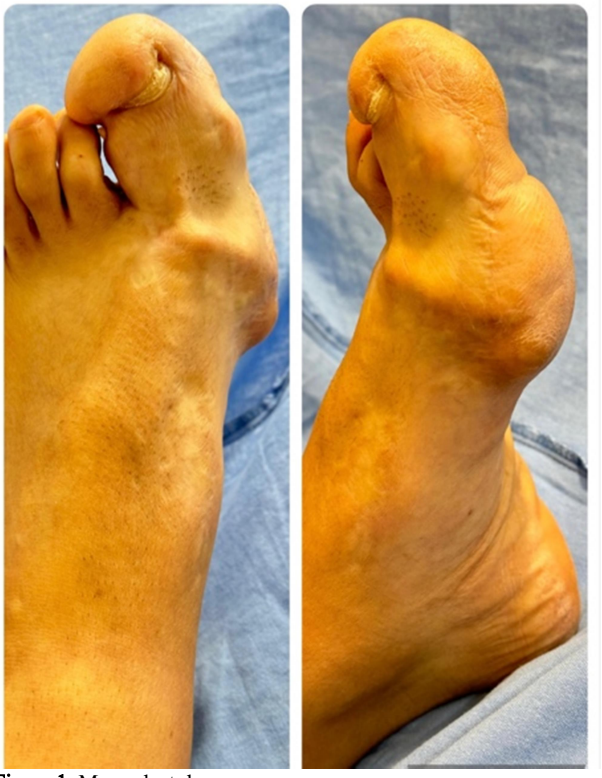

Figure 1. Macrodactyly

Case report

A 38-year-old female patient with no history of significant diseases, with a diagnosis of progressive macrodactyly of the left 1st toe, which was evident from 3 months of age with slow growth. She has a history of 2 previous corrective surgeries, the first at 12 years of age, with a reintervention 8 months later in which 2 nails were used for rectification.

Currently with a recurrence, after her last pregnancy which was 7 years ago, she presented growth in both the metatarsal and the first toe, completely asymptomatic. She comes to the clinic due to pain when wearing shoes, denies any limitation on walking, and is neurovascularly preserved. (Figure 1)

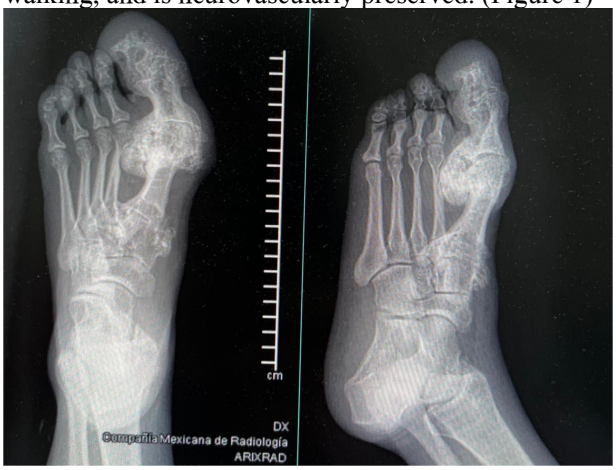

Figure 2. X-ray of the left foot with calcified nodules

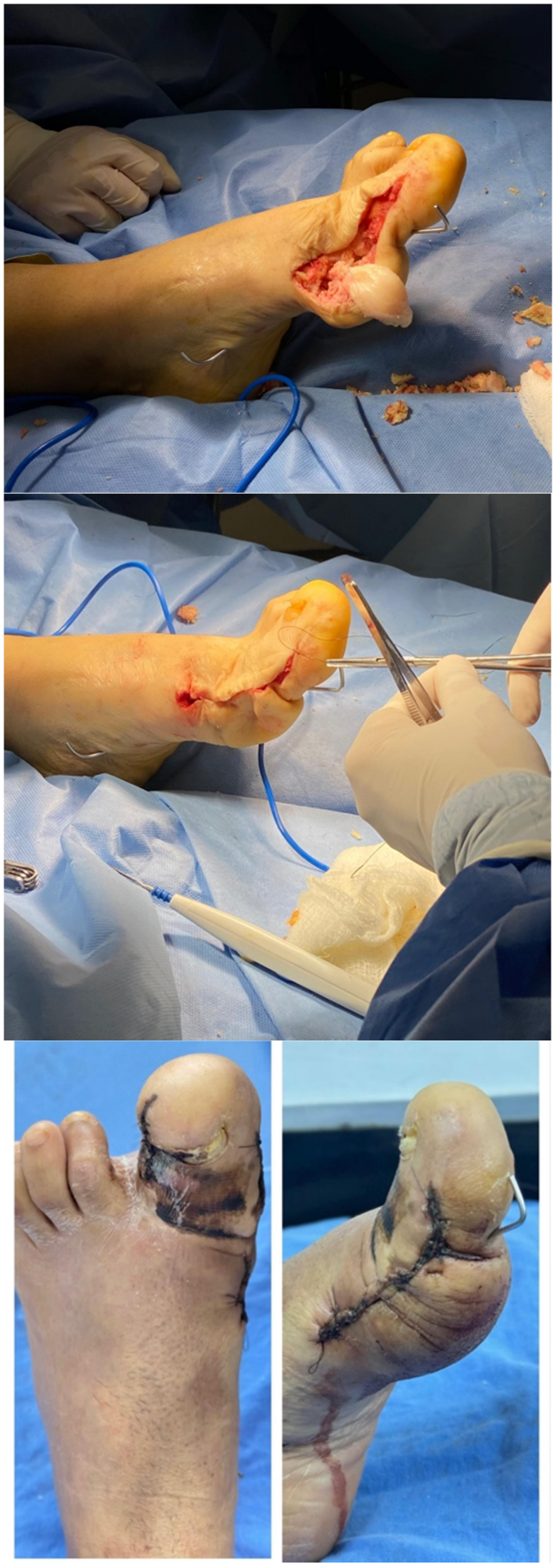

Figure 3. *Above.* Osteotomy and fixation . *Middle.* Closure. *Below.* Patient 8 days post-surgery.

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An x-ray of the left foot was performed with the presence of a tumor in the middle and distal phalanx of the left foot with multiple calcified nodules with spiculated edges, for which a scintigraphy was requested to rule out tumor origin. (Figure 2)

Subsequently, a CT scan with bone reconstruction is requested for surgery planning by the Plastic and Reconstructive Surgery service in conjunction with Traumatology and Orthopedics.

Reduction of the 1st finger is performed through osteotomy of the phalanx and fixation with Kirschner wires by the traumatologists and orthopedists and the soft tissue remodeling and closure by the plastic surgeons, resulting in the surgical event without complications. (Figure 3)

In post-surgical review, we found a wound without dehiscence and with no signs of infection, and most importantly with a decrease in the size of the finger. (Figure 3)

Discussion

Macrodactyly is a rare congenital abnormality, characterized by overgrowth of the bones and soft tissues of the fingers. The metatarsals, tendons, and vessels in the toes are usually not affected. Wu et al studied 73 cases (12 static and 61 progressive) and found that the most commonly affected finger was the second[2].

The etiology and pathogenesis are not clearly defined or understood; However, there are multiple hypotheses, among which lipomatous degeneration, abnormal development of somatic cells during growth, alteration of fetal circulation during intrauterine development, segmentation errors and dysfunction of autonomic nerves, among others. Due to the low frequency of this disease, these associations have not been demonstrated in any series; however, there is a high association of overgrowths with some musculoskeletal syndromes. Radiographic studies show soft tissue hypertrophy and overgrowth of the phalanges and metatarsals. The MRI indicates an overgrowth of non-capsulated fat and fatty infiltration that may lie towards the muscle[4].

In macrodactyly, 90% of cases are unilateral and the involvement of multiple fingers is three times more common than that of a single finger. All structures are hypertrophied, especially the digital nerves and fatty components. The tendons and vessels are only slightly larger, without any notable pathological findings[3].

Two types of growth patterns are classically described: static and progressive, and the distinction is made on clinical grounds. Both types are present at birth, in the static type the affected parts grow proportionally with the child, while in the progressive type there will be disproportionate growth, which becomes evident at the age of 2 to 3 years. Surgery with aggressive soft tissue and bone reduction is necessary earlier. In a small number of patients, a third pattern of unrestricted exaggerated growth is seen in which the affected parts are so large that they usually require early amputations, as attempts at initial aggressive surgical reductions usually fail[3].

Treatment is very difficult due to the progressive and diffuse nature of this nosological entity that prevents complete correction to normality. The indications for surgery are aesthetic and functional[1].

Most patients require multiple surgical interventions during childhood and in a high percentage of them, the result is an unsightly and dysfunctional finger[1].

Among the most frequently used techniques is volume reduction surgery or dermolipectomy, which aims to remove the greatest amount of skin, subcutaneous tissue and osteophytes as long as it is possible to close the edges of the incision without tension[3].

Debulking and reshaping procedures can be performed at any time. A disproportionately large finger should not be left untreated during school age, since adjustment and psychological problems occur more in this group[3].

In some cases, several volume reduction procedures are necessary due to progression, recurrence or persistence of overgrowth of the soft tissues of the finger. If possible, surgeries should be performed near the end of growth, unless exaggerated overgrowth of the finger requires early surgery[3].

Amputation of the affected rays is the method with the best results, when carried out at ages between three and six years. Chan et al mention that ray amputation is indicated when the disease is unilateral and the intermetatarsal angle is 10 degrees or greater compared to the contralateral angle, while Dedrick and Kling recommend this amputation when the affected toe is two standard deviations above the length of the contralateral[4].

The decision to perform panepiphysiodesis on the big toe is made when the size reaches the length of the homologous toe of the parent of the same sex. There are some other techniques described to improve the cosmetic appearance of the foot and prevent overgrowth of the toes; which include reduction osteotomies, phalangectomies and epiphysiodesis; all of them with variable results and some not favorable[4]. According to the evidence, aggressive surgical management is the most useful and recommended in the case of macrodactyly in the feet. Amputation of overgrown fingers from the base of the metacarpal represents to date the most viable option with the least possibility of recurrence. The functional result takes precedence over the cosmetic one, and the objective

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assessment scale (PODCI) demonstrates that the final result and prognosis in function with this treatment surpass the semiconservative treatment resecting only the overgrown soft tissue[4].

Conclusions

Macrodactyly is a rare disease with a benign course that represents a challenge for surgeons due to its difficult management and high recurrence rate, which leads to multiple surgical procedures with variable results. The goal of surgery is to have an aesthetic, functional and pain-free foot as the final result. It also carries an important psychological factor for both the patient and the family.

Our patient has a history of 2 previous procedures for resection of fatty and bone tissue which were not definitive for the cure of the condition since the tissue overgrowth in the foot continued, leading to the need for another surgical procedure for which we approached her.

Amputation as an early definitive treatment avoids multiple surgeries to save an unsightly toe with few functions, but in our particular case, despite presenting recurrences, amputation was not considered since the big toe is essential for balance and gait propulsion, plus she is a young patient and did not want that treatment option.

Ideally, each case should be personalized to determine the appropriate time to begin surgical procedures and the most optimal surgical plan to be carried out in order to achieve better functionality and aesthetics of the affected finger.

Conflicts of interest

The authors have no conflicts of interest to declare.

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