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CASE REPORT OPEN ACCESS

MACRODYSTROPHIA LIPOMATOSA: A CASE REPORT

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ABSTRACT

Lipomatous macrositrophy, also known as macrodactyly, is a rare congenital condition of overgrowth of bone structures. Its diagnosis requires a close relationship with clinical, histopathological and imaging data, in order to rule out other possible causes of macrodactyly, in addition to tumors and malformations. The present report describes a 31-year-old patient, with continuous growth of the 3rd and 4th fingers of the left hand, alleging nervous, motor changes and a strong influence on the biopsychosocial aspect. The patient underwent hand radiography, hand computed tomography and skin biopsy, covering all aspects necessary for the final diagnosis of lipomatous macrodystrophy.

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INTRODUCTION

Macrodactyly, also known as digital gigantism, corresponds to a rare congenital abnormality that encompasses approximately 1% of all innate anomalies (Batista *et al*, 2008). It is characterized by the disproportionate enlargement of the fingers or toes, usually found in the true isolated form. However, it is essential to distinguish it from tumors and other malformations, such as congenital lymphedema, lipomas, Ollier's disease (multiple endonchondromas), Klippel-Trenaunay-Weber syndrome, osteoid osteoma (Monteiro *et al*, 1998). Initially, digital gigantism was described by Klein, in 1821 and affects both sexes, with a predilection for the male sex, predominantly unilateral, with rare cases of bilaterality described (Klein, 1824; Monteiro *et al*, 1998; Mehra *et al*, 2017) . In addition, it presents two main forms: static and progressive. The first occurs since birth, when the proportional overgrowth of a finger occurs and which keeps its dimension proportionally static in relation to the others during the period of skeletal maturation. The second form occurs when the affected finger may not be as wide during birth and later deform with skeletal maturation (Monteiro *et al*, 1998; Mehra *et al*, 2017; Watt and Chung, 2004; Celebi *et al*, 2015).

It is worth mentioning that macrodactyly can be known by its synonyms: lipomatous macrodystrophy, fibrolipomatous macrodactyly, localized gigantism and megalodactyly (Monteiro *et al*, 1998). Such pathology entails psychological and functional sequelae that accompany the individual from the moment they settle in - at birth, if it is the static form and at the time when the increase is impossible not to be noticed, that is, in the progressive form (Batista *et al*, 2008).

CASE REPORT

Male patient, 31 years old, resident in the interior of the state of Pará, is admitted to the Dermatology outpatient clinic, complaining of a progressive growth of the 3rd and 4th left fingers (figures 1 and 2), 5 years ago. As associated symptoms, the patient reported pain and local paresthesia, in addition to having difficulty in carrying out manual work and social isolation caused by hand deformity. He denied any history of trauma, previous treatment or other systemic symptoms. The patient underwent radiography of the left hand, which attested to increased bone and soft tissue dimensions of the 3rd and 4th fingers (figure 3). The examination was complemented with computed tomography of the left hand, which revealed a

volumetric increase of the 3rd and 4th fingers, due to the presence of anomalous tissue predominantly adipose from the soft tissues, associated with areas of cortical bone hypertrophy close to the proximal and distal interphalangeal joints. Thus, the findings favor as a diagnostic hypothesis of lipomatous Macrodystrophy, with type 1 neurofibromatosis among the differential diagnoses (Figure 4).

Figure 1. Lipomatous macrodystrophy with disparity in size of the 3rd and 4th fingers on the posterior face

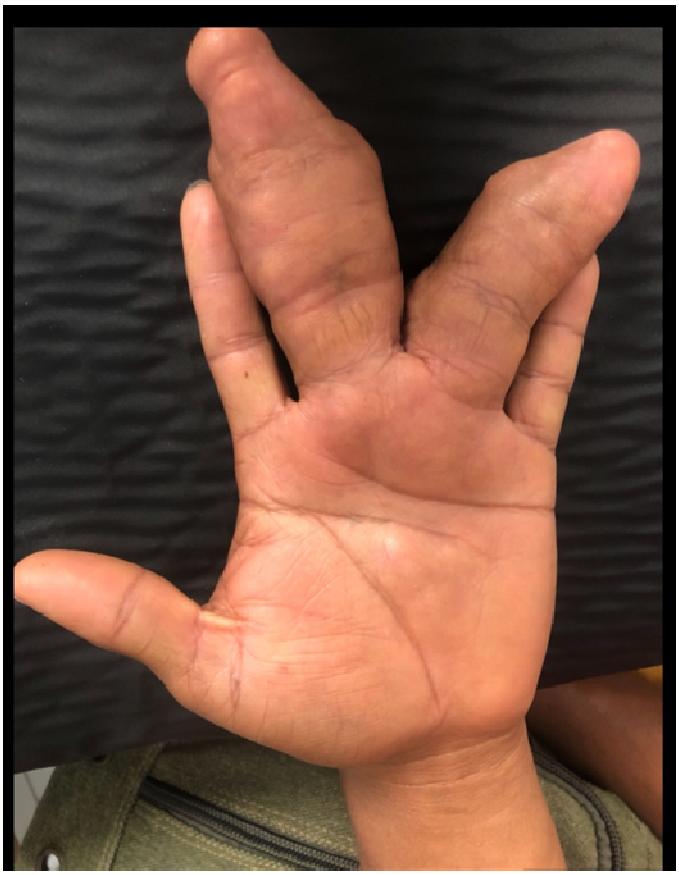

Figure 2. Lipomatous macrodystrophy with disparity in size of the 3rd and 4th fingers on the anterior aspect of the left hand

The patient was then submitted to skin biopsy, which confirmed the presence of mature adipose tissue proliferated in lobes and permeated by fibrillar fibrous tissue, separated by discrete fibrous septa. In addition, there was an absence of vascular and neural proliferations, a fact that precludes other causes of macrodactyly (Figure 5).

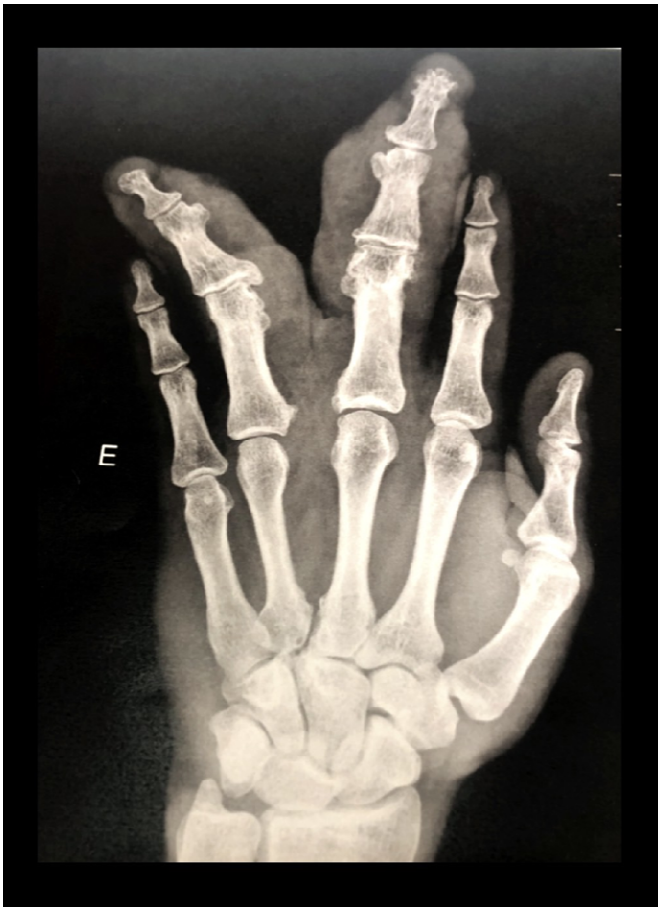

Figure 3. Radiography of the left hand with increased bone and soft tissue dimensions of the 3rd and 4th fingers

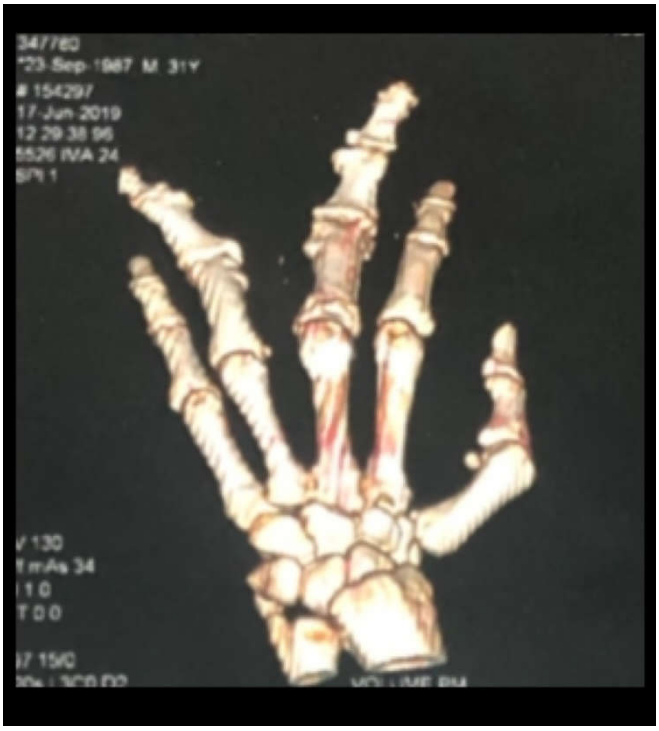

Figure 4. Computed tomography of the left hand with cortical bone hypertrophy close to the proximal and distal interphalangeal joints

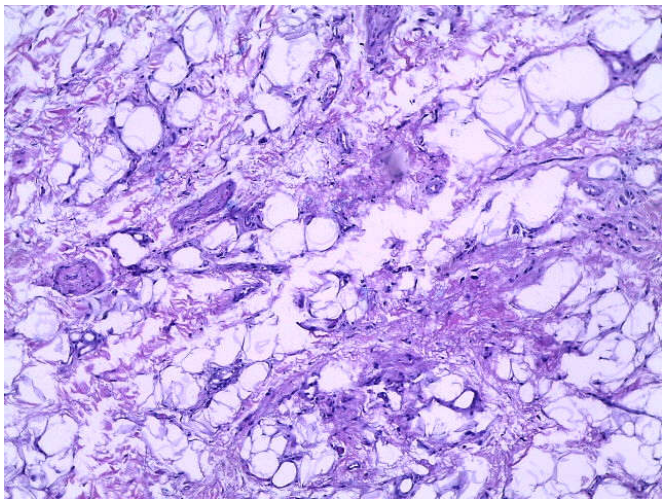

Figure 5. Microscopy of the skin biopsy revealing the presence of mature adipose tissue interspersed with fibrillar fibrous tissue and fibrous septa, compatible with lipomatous macrodystrophy

In this context, after establishing the diagnosis of progressive macrodactyly, clinically and radiologically, the patient received a prescription for analgesia and was referred to specialized orthopedic care to assess therapeutic possibilities.

DISCUSSION

Disorders that involve overgrowth of the hands are contained in a group of diseases that have a similar phenotype, making the diagnosis complex and challenging. In the past, many of these conditions were called hamartomas. Thus, the term macrodactyly was later incorporated to cover a group of diseases where there is an increase in the fingers, including: neurofibromatosis, Proteus syndrome, hemihypertrophy, Maffucci syndrome, CLOVE syndrome, lipomatous macrodystrophy, Parkes Weber syndrome and Klippel-Trenaunay syndrome. Such pathologies have their own clinical characteristics that help in the diagnosis: presence of hemodynamic changes, vascular anomalies, other associated anomalies, nerve involvement, coagulopathies, among others (Carty *et al*, 2009). Macrodactyly or lipomatous macrodystrophy is unilateral, in 90% of cases, and generally affects more than one finger, with all of its structures affected by enlargement. Meanwhile, digital involvement is the most frequent, although the deformity can extend to the palm and / or forearm in 7% and 4% of cases, respectively. In such cases, overgrowth leads to angular deviation of the affected finger, also known as clinodactyly; also syndactyly, polydactyly, brachydactyly (Boves, 1977; Mehra *et al*, 2017; Watt and Chung, 2004; Celebi *et al*, 2015). The most affected fingers are the thumb, index and middle. It is also worth mentioning that the most probable hypotheses for macrodactyly overgrowth are: abnormal nervous supply; abnormal blood supply and abnormal hormonal mechanism (Inglis, 1950). Among the main nerves affected, there is the median nerve - in the upper extremities - and the plantar nerve - in the lower extremities (Mehra *et al*, 2017; Watt and Chung, 2004; Celebi *et al*, 2015).

This characteristic is compatible with the present report, in which the 3rd and 4th fingers were affected. It is also worth mentioning that the repercussions involve aesthetic and mechanical causes. Thus, the patient may suffer interferences, problems and prejudices in daily life, with serious biopsychosocial repercussions. In addition, this increase in structures can cause a reduction in function, in addition to slowness in the electrical conduction of the corresponding nerves, whether of motor or sensory function (Mehra, 2017; Watt and Chung, 2004; Celebi *et al*, 2015). Regarding treatment, it must be different for each case. The type of macrodactyly, the speed of disease progression, the fingers involved and the patient's age must be taken into account (Monteiro, 1998). With the treatment, the objective is to provide a hand with the ability to clamp and grasp objects, in addition to being aesthetically acceptable (Bisneto, 2013). Given this context, several techniques, which combine osteotomies, epiphysiodeses, reduction of soft tissues - especially fatty tissue - tendon shortening and neurectomies, aim to prevent hypergrowth and reconstruct the appearance of the finger (Bisneto, 2013). Finally, the importance of the association of detailed anamnesis with physical examination and images, as well as histopathological confirmation and the study of the different differential diagnoses for the early identification and treatment of this rare disease is emphasized.

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