

On a case of "Macrodystrophia Lipomatosa"

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Abstract. – Macrodystrophia Lipomatosa is a congenital malformation of rare finding and unknown pathogenic mechanism. The pathology is mainly characterized by the interest of lower extremities and peculiar macroscopic feature is the presence of hypertrophic fibro-adipose tissue. In our experience the localization to the upper extremities and the presence of uncommon clinical signs show the importance of instrumental investigation: MRI and CT for an accurate examination and to exclude other types of localized gigantism.

Key Words:

Macrodystrophia lipomatosa, RMI, Hypertrophic fibro-adipose tissue.

Introduction

ML is a congenital malformation of rare finding first discovered by Kelikian in 1925 (having discovered 20 cases in the lower extremities). Only a few years later Ranavat and colleagues found the same pathology in the upper extremities.

Successive years found clinical cases on the decline and only in 1967 was Barsky able to define certain peculiar characteristics (Table 1, Figure 1).

1. Localized gigantism in an uniform fashion of the digits¹.
2. Not hereditary.
3. Predominantly in the lower extremities.
4. Rare associations with vascular diseases.
5. A pathology affecting the mediane nerve of the upper extremities, and the posterior tibial nerve of the lower extremities, specifically the 2nd and 3rd digits.
6. A classic sign is the presence of the hypertrophic fibro-adipose tissue.

Of all the theories trying to describe the etiopathogenesis of ML the two accepted

seem to be: Disembriogenesis and the nervous theory.

The first, requires the action of one or more noxae patogene during the intrauterine development of the mesodermic structure of the extremities.

The second theory, the one most accepted by most of the authors, is that of neurotrophic disturbances, hypothesizing a direct influence of the nervous structures on the distrectual growth.

Still today however, the pathogenic mechanism remains obscure and confined within the limits of supposition^{1,2,3}.

Clinical Event

The event brought up to our observation concerns a 40 years old patient (female) who was admitted in our section with a diagnosis of carpal syndrome of the right wrist.

The anamnestic investigation showed absence of familiarity and the patient referred the presence of digital deformity localized to the hands in association with diffuse angiomas since the birth^{1,2}.

Table I. The Macrodystrophia Lipomatosa.

The pathology is characterized by an elective localization of lower extremities, rarely upper extremities
Rare bilaterality
Rare association with vascular diseases and absence of artero-venous fistula
The Macrodystrophia Lipomatosa is a pathology affecting the median nerve of upper extremities, and the posterior tibial nerve of lower extremities, specifically 2 nd and 3 rd digits



Figure 1.

The above described deformity gradually developed and it came to an end at puberty.

At the age of 35, without paying attention to the aesthetic appearance, the patient started to feel an acute pain of the carpal nerve.

As time went on, these symptoms became worse causing her a greater degree of limitation.

The objective examination revealed the presence of macrodactyly and clinodactyly affecting three fingers of the right hand (elective interest of the first, second and third fingers of the hand) with increased volume in the forearm, and diffuse angiomas of the trunk, neck and forearm.

The presence of objective signs common to other pathologies made it impossible to for-



Figure 3.

mulate a diagnosis of ML, and suggested other forms of localized gigantism (Klippel-Trenaunay-Weber syndrome and Neurofibromatosis) (Figures 2-5).

As a matter of fact, common signs to several forms of gigantism are¹⁻³:

1. Presence of angiomas
2. Gigantism localized all-over the limb
3. Bilaterality
4. Frequent pathology of the median nerve (specific areas, tributaries of the median nerve, are interested).

At the same time, traditional instrumental investigations didn't help to make a diagnosis not revealing any valuable data. However, an accurate radiologic investigation revealed^{2,4,5}:



Figure 2.



Figure 4.

1. Osteophytes on the specific phalanges
2. Abnormal growth of cortical bones
3. Increased volume of bones
4. Clinodactylia
5. Clinical signs of degenerative artropaty and abnormal growth of soft tissues.

(Tables—2-3; Figures 6-8)

No artero-venous fistula were found after an angiografic investigation.

Nevertheless a different form of SKTW, (meaning the Parke Weber syndrome), could not be excluded, this syndrome in fact is mainly characterized by the absence of the artero-venous fistula³.

A conclusive diagnosis was possible only after a CT (tomoscan s.r. 7000 Philips), together with MRI (girosan 0'5 Philips) (Table 4; Figures 9a-9b).

The results obtained showed the presence of *hypertrophic fibro-adipose tissue (peculiar characteristic of this pathology)*⁴⁻⁵.



Figure 5.



Figure 6.

Conclusions

The case reported represents only a contribute in the uncommonity of the pathology (Macro dystrophia lipomatosa).

The presence of uncommon clinical signs of this physical abnormality shows the importance of instrumental investigations CT and MRI for a correct diagnosis and to exclude other types of localized gygantism^{6,7}.

Above all RMI represents the methodic of choice able to characterize the soft tissues' alteration. Particularly RMI shows the clear presence of abnormal soft tissue and the fibro-adipose infiltration of the muscles.

Table II. Radiographic alteration of Macro dystrophia Lipomatosa.

Radiographic alteration of ML are an abnormal growth of soft tissues, an abnormal growth of bones, clinodactylia and signs of degenerative artropathy

Table III.

In the case presented there is an elective interest of 2nd right hand's finger (metacarpo and phalanges) with clinodactylia, camptodactylia and arthropathy

Table IV. CT and MRI alterations of Macro dystrophia Lipomatosa.

CT and MRI investigations shows the peculiar characteristics of this pathology "an abnormal growth of soft fibro-adipose tissue that extends itself into the surrounding muscles without infiltration".



Figure 7.

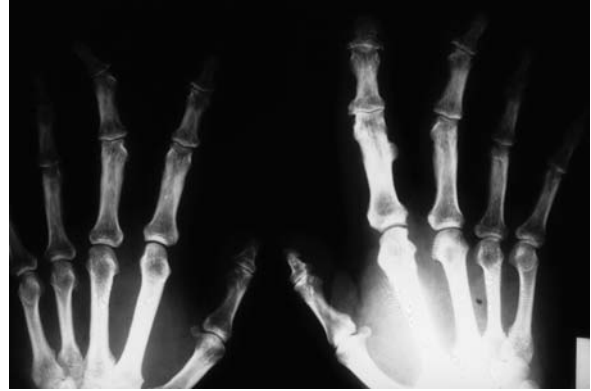


Figure 8.

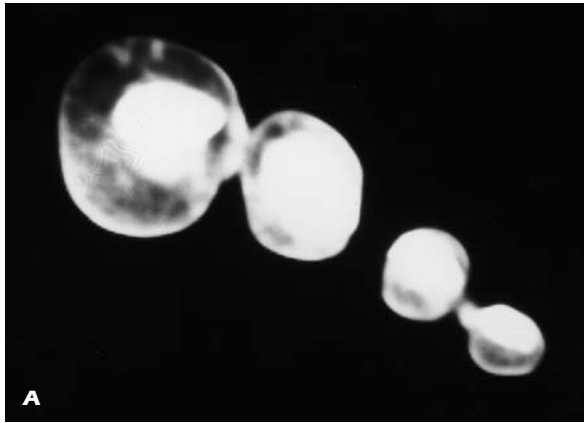
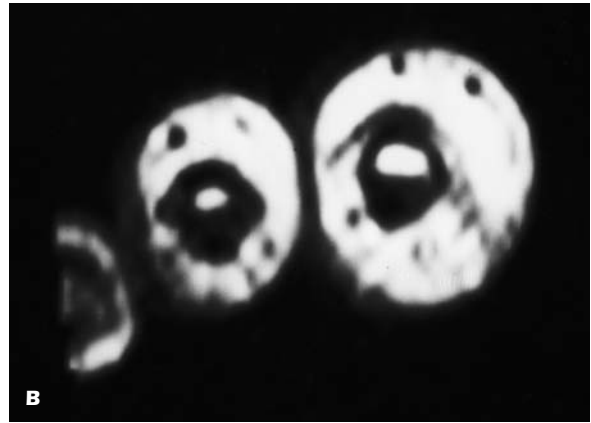


Figure 9.



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