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Invasive macrodystrophia lipomatosa of the hand

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Abstract. – OBJECTIVE: The Authors point out the interest of differential diagnosis and conservative surgical treatment of a rare case of digital and ulnar side of the hand gigantism, with massive fatty infiltration of soft tissues and a neurovascular bundle, to be included into Macrodystrophia Lipomatosa with fibrolipomatous hamartomata.

PATIENTS AND METHODS: Excision of the mass included 4th ray disarticulation (the fifth had been amputated several years ago) as well as microsurgical external and internal neurolysis of the ulnar nerve, the ulnar neurovascular bundle being exposed and covered with dermal substitute INTEGRA™, with a good result both from a cosmetic and functional point of view at three year follow up.

RESULTS: This is the first report of INTEGRA™ covering of a neurovascular bundle.

Samples taken from the dermal substitute matrix interface at day 6 and day 25 were examined with transmission Electron Microscopy: a newly formed tissue, rich in precursor cells, was detected.

CONCLUSIONS: Conservative surgery, requiring particular microsurgical skills and use of dermal templates, plays an outstanding role in treatment of these rare but aggressive soft tissues masses of the upper limb.

Key Words:
Macrodystrophia lipomatosa, Hamartoma, Upper extremity, Nerve, Neurolysis, Neurovascular bundle, Dermal regeneration template.

Case Report

A 48 year-old, manual worker, presented to our Unit complaining of a soft, painful swelling of the ulnar side of his left hand. When he was 12, he had undergone fifth ray amputation because of digital gigantism. Mother's pregnancy as well as family history did not show any remarkable element.

Physical examination showed a bulky mass of the whole hypothenar region with fourth ray macrodactyly, the wrist being also involved, with some soft nodules palpable along the ulnar side. He experienced neither pain nor hyperesthesia of his thumb, index and middle fingers, was able to grasp, but the fourth finger was stiff and itching and paraesthesias were produced by simple touch with the ulnar side of the hand and the range of motion of the left wrist was strongly restricted.

Radiographs revealed marked radiolucent areas, suggesting the presence of fat, together with a larger second phalanx of the fourth digit with exostosis-like osseous overgrowth of its distal end (Figure 1).

Excision of the infiltrating mass included 4th ray remnant disarticulation at the metacarpophalangeal (MP) joint and opening of the Guyon tunnel and carpal tunnel with extensive neurolysis of both the median and the ulnar nerve, which needed to be freed from an important fatty infiltration.

Neurolysis from fat was performed up the origin of the common trunks, only the most ulnar of which demonstrated a typical serpentine aspect; at the end of excision, the exposed ulnar neurovascular bundle was covered by INTEGRA™ (Figure 2). Samples for pathology were taken at day 6 and 25.

A split thickness skin graft was applied 25 days later; the patient recovered his left hand function, gaining satisfying functional and aesthetic result.

A Transmission Electron Microscopy study showed cell/neomatrix interface and invasion by newly-formed cell precursors (Figure 3).

Concerning with sensation recovery, we used a modification of Mackinnon and Dellon Sensory recovery scale based on both static and moving 2-point discrimination[1] and experienced poor results (S0-1) in the first six months, followed by a slow recovery in the following six months, leading to S2 (at least protective sensitivity) one year after surgery.

Both aesthetical and functional outcome have revealed satisfactory at three year follow up (Figure 4).

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Figure 1. Clinical and radiological views of the affected hand.

Discussion

Macrodystrophia lipomatosa (ML) is a rare nonhereditary form of localized gigantism characterizing by osseous and fibroadipose overgrowth as main features, classically affecting hand or foot in a median or plantar nerve distribution. Favoured sites of involvement are the second and the third digits in both the upper and the lower extremities, the latter being more commonly involved. Fat deposition could be revealed within the nerve sheath, bone marrow, periosteum, muscles and subcutaneous tissues[1-3].

Various hypotheses have been proposed regarding the pathogenesis of ML, including lipomatous degeneration, fetal circulation abnormality, and damage of extremity bud and errors in the segmentation in intrauterine life and hypertrophy of the concerned nerve.

Association with macrodactyly and other hamartomata can be explained by anomalous nervous and paracrine pathways generated in the affected limb; anomalous fat and fibrous infiltration of the nerve sheaths could promote an uninhibited production of growth factors, and determine gigantism of the relative segment of innervation[1].

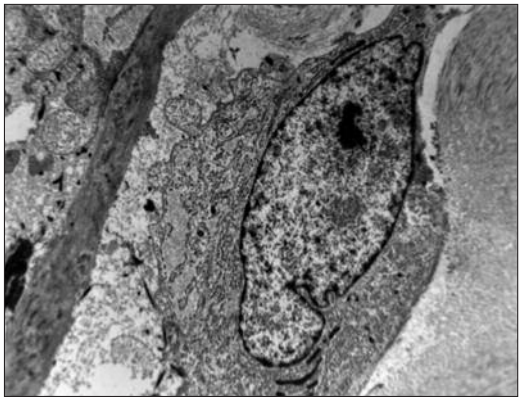

Figure 3. A cell-rich newly formed tissue with active precursors is detected; large-nucleated precursor cells make their appearance (transmission electron microscopy (8000x).

Anyway, the gamut of nomenclature by different authors led to a considerable confusion by time: the first mention of the term *macrodystrophia lipomatosa* by Feriz in 1925 was followed by the description of a progressive form of local gigantism by Barsky in 1967[4].

Similarly, the above mentioned *fibrolipomatous hamartoma* represents another eterogeneous group of lesions which produces digital overgrowth, usually presenting as an isolated nerve lesion and associated with intramuscular fat deposition, with no involvement of periosteum nor bony changes[5].

In ML, in fact, the periosteum can be seen studded with small nodules of chondroblasts, osteoblasts and osteoclasts, which become larger and numerous toward the distal ends of the phalanges, producing elongation and broadening up to a "mushroom-like" appearance. Secondary osteoarthritic changes, as joint space narrowing, subchondral cysts and large osteophytes can also be observed[6].

Patients suffering from ML usually complain both cosmetic and mechanical problems.

Figure 2. The ulnar neurovascular bundle has been covered with dermal template INTEGRA™.

Figure 4. Satisfactory outcome after surgery (three-year follow-up visit): palmar view.

Usually, tissue overgrowth in the volar aspect produces dorsal deviation of the affected part, with an important impairment in normal daily activities.

Degenerative joint changes, together with compression of adjacent nerves and vessels, commonly produce carpal tunnel syndrome and other entrapment conditions. Electroneurography and nerve conduction velocity tests reveal slowed distal motor and sensory transmission, local/segmental conduction block or slowing of the peripheral nerves at the entrapment sites[5,6].

Radiographic findings of this condition are usually pathognomonic and are enough to make diagnosis[1,5,7]. Typical x-ray findings include excessive growth of soft tissue as well as osseous tissue, presence of radiolucent areas due to the adipose tissue infiltration, and degenerative joint disease.

Excessive growth of the bone within the area innervated by nerve and fat tissue proliferation within the muscle fibres are the characteristic findings detected on CT scan. The excessive fat seen in ML is not encapsulated and MRI can easily demonstrate the fatty infiltration of the muscles[5,7]. Fatty infiltration of nerves is a typical finding from MRI, as well as deserved by pathological observation[3,7] such as we also documented.

Differential diagnosis should include other possible causes as Proteus syndrome, Ollier disease, neurofibromatosis and several lymphatic and vascular lesions, as in Klippel-Trenaunay-Weber syndrome, Maffucci syndrome, lymphangiomatosis and haemangiomatosis[6,7]. Absence of familial occurrence and lack of any cutaneous or systemic manifestation makes many of these diagnoses unlikely[1,3,8].

The main target of the surgical intervention is represented by improving cosmetic appearance as well as preserving neurologic function.

The way of treatment depends on patient's age. Pediatric patients are submitted to early surgery (microscopic dissection of tumor from nerve sheath) to limit further operations, as they are expected to develop synaptic plasticity, nervous regeneration potential and neural crossover, usually recovering nerve function. Unfortunately, ML patients undergo numerous interventions during lifetime, because of serial presenting of new masses and recurrences. In adult patients functional neural impairment, such as sensitive and sweating loss, claims for more aggressive surgery with extensive neurolysis and only rare cases can be conservatively treated by volar carpal ligament release only[5].

Our case demanded an important excision with segmental disarticulation and extensive neurolysis of both median and ulnar nerve. INTEGRA™ was an interesting solution to cover an exposed neurovascular bundle, as this indication has not yet been reported.

We were able to compare our experimental data of enveloping a neurovascular bundle with an artero-venous loop into an INTEGRA™ sheet[9], observing that both angiogenesis and neural regeneration were enhanced, leading to a soft repair with a protective sensitivity.

Although it has been assumed that ML regress to a static lesion after completion of growth, our case shows that some lesions tend to slowly enlarge in the adulthood or to recur[3,9].

The patient has been invited to a year-by-year follow-up.

Conflict of Interest

The Authors declare that there are no conflicts of interest.

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