

Lipofibromatous Hamartoma of the Median Nerve in Patients with Macroductyly: Diagnosis and Treatment of a Rare Disease Causing Carpal Tunnel Syndrome

Dr. Milandeep¹, Dr. Gajendra Pandit², Prof. Dr. Darshan Singh Sidhu³

Abstract: *Background: Lipofibromatous hamartoma or Macrodystrophia lipomatosa is a very rare benign peripheral nerve tumor most commonly affecting median nerve in young adults. It is a form of local gigantism characterised by disproportionate increase of adipose tissue and mesenchymal elements in neural distribution. Case: An 18yr old female patient with complaints of macroductyly without any neurological symptoms was diagnosed with lipofibromatous hamartoma based on the operative findings which included enlargement of median nerve and neurovascular bundle of fingers. This enlargement was due to fibro-fatty tissue infiltration in nerves. Result: The treatment involves carpal tunnel release to decompress the median nerve, excision of the redundant tissue around the nerves and debulking of soft tissue component. Discussion: The predominant association of median nerve in this condition is unexplainable. The associations of Lipofibromatous hamartoma of median nerve are macroductyly or macroductyilia fibrolipomatosis, ectopic calcification of soft tissues, bone exostoses, subcutaneous lipomas and vascular tumors.*

Keywords: Lipofibromatous hamartoma , carpal tunnel syndrome, macroductyly, median nerve

1. Introduction

Lipofibromatoushamartoma is a rare and slow growing benign fibro-fatty tumor¹. It is characterized by the proliferation of mature adipocytes within the epineurium and the perineurium of the peripheral nerves. In the upper extremity, it most commonly affects the median nerve².

It commonly affects the volar aspect of the hands, wrists and forearms of young adults. Most patients present either early with macroductyly or later with a forearm mass lesion or symptoms consistent with compressive neuropathy of the involved nerve. Median nerve involvement commonly leads to pain, numbness, paresthesia and carpal tunnel syndrome³. Most respond to conservative management with surgical exploration, biopsy and carpal tunnel release to decompress the nerve. Correct diagnosis of this uncommon lesion is important as surgical excision of the lesion may lead to loss of neurological function.

2. Case Report

A 18 year old female came to our surgical OPD with complaints of swelling over the right middle and index finger since birth, curvature of the right middle finger and restricted movements of the involved fingers. Her parents noticed the steady enlargement of her right middle and index fingers since childhood and had been treating it in a local hospital without any significant improvement. Slowly the movements of the involved fingers got restricted and the enlargement lead to the curvature of the middle finger medially. There were no sensory disturbances. All other fingers were normal for age and all movements intact. No history of numbness, paresthesia and burning sensation. No history of fever and ulceration or discharge from the involved fingers. Her family and natal history did not reveal anything significant.

The findings in this patient included

- 1) Macroductyly of Rt middle and index fingers
- 2) Middle finger curvature towards medial side
- 3) Thenar eminence atrophy
- 4) Phalen's test and Tinel's sign positive

All routine blood investigations were within normal limits. Ultrasound colour Doppler was done which also turned out to be normal. Auto antibody assay was also negative.



X-ray



The operative findings in this patient turned out to be Median nerve hypertrophy with fibro-fatty tissue leading to carpal tunnel syndrome along with distal neuro-vascular bundle hypertrophy.

Carpal tunnel release was done to decompress the median nerve, followed by debulking of overgrown soft tissue in the distal fingers. Finally to correct the curvature the distal phalanx of middle finger was osteotomied.

We planned and took the patient for Carpal Tunnel Release and debulking of Rt Index fingers.



During Operation Hypertrophy with Fibro-Fatty Tissue of Median Nerve



After 1 Month of Operation

3. Discussion

Etiology and Involvement:

Lipofibromatous hamartoma (LFH) of the nerve is an uncommon neural non-neoplastic lesion demonstrating

gross enlargement of a nerve caused by epineural and perineural proliferation of fibrofatty tissue(5). Less than 100 cases of LFH affecting the median nerve(6) have been documented, of which about one-fourth were associated with true macrodactyly in the territory of the nerve distribution,

predominantly in females. The macrodactyly can be present at birth or can be diagnosed during early childhood. There is no report of familial history or chromosomal abnormality in this condition.

Many names have been used to describe LFH in the past including fibrolipomatoushamartoma, fibrolipoma, lipofibroma, fibrofatty proliferation, microdystrophialipomatosa, intraneurallipoma, perineurolipoma, fatty infiltration of the nerve, fatty or fibrous neoplasm of nerve and lipomatosis of nerve.(7) In 1969, the term lipofibromatoushamartoma was introduced by Johnson and Bonfiglio and remains the most accurate description of this condition (8)

Macrodactyly

True macrodactyly is characterized by an increase in size of all elements or structures within the digit or digits. There are two types of true macrodactyly. In the first type, the enlargement is present at birth and does not increase disproportionately with growth. This is the static type, or *macroductylicaongenita*. In the second type, there is disproportionate growth of the involved digits. They grow much more rapidly than normal digits and frequently deviate to one side. Another common but very important complication of this type is overgrowth of fatty tissue in the palm and proximal part of the affected limb. This is the progressive type of macrodactyly, or *macroductyphia lipomatosa progressiva*.

Gross and Pathological findings

Neural fibrolipoma is an orange- yellow, fusiform, sausage like or ropelike enlargement of the nerve. Involved nerves may be markedly increased in length and diameter. The lesion is described as a hamartoma because of the overgrowth of the normal connective tissue constituents: fat and fibrous tissue. Histo-pathological examination shows infiltration of the epineurium by fibrofatty tissue. This separates and compresses the nerve fascicles and causes atrophy of the neural elements, often accompanied by extensive perineural fibrosis.(9)

Management

The surgical management of LFH of the median nerve remains controversial. Decompression of the carpal tunnel, debulking of the fibrofatty sheath, microsurgical dissection of the neural elements, and excision of the involved nerve with or without grafting are recommended. Tumors that infiltrate the nerve are difficult to extract and hard to access. For these patients, performing an epineurotomy and intraneural dissection have seen good results.(10)

The lesion can be debulked extensively to improve the functional status. Extensive microsurgical intraneural dissection can lead to significant ischemic complications. Aggressive surgery should be only be used if conservative management fails or when the benefits of surgery outweigh the risks.

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