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Review Article

A case of macrodystrophia lipomatosa of 5th digit of hand with literature review

Vijayraj Patil*[1], Abhishek Vijayakumar[1], Avinash Vijayakumar[2]

[1]*Bangalore Medical College and Research Institute, Fort, K.R. Road, Bangalore - 560 002, Karnataka, India*
[2]*Department of Radiology, Banaras Hindu University India.*

***Correspondence Info:**
Dr. Vijayraj Patil,
Bangalore Medical College and Research Institute,
Fort, K.R. Road, Bangalore - 560 002, Karnataka, India
E-mail: vijay.495.495@gmail.com

Abstract

Macrodystrophia lipomatosa (ML), a rare congenital disorder, is considered by some to be a localized form of Proteus syndrome. A 12year old girl presented with progressive enlargement of little finger of right hand since birth. Magnetic resonance imaging showed unencapsulated excessive fibroadipose tissue infiltrating interosseous muscles. Partial debulking of the lipomatous tissues was performed. Microscopic examination of the specimen revealed fat deposits, ligamentous, and neural tissue elements in normal structure. We report a case of atypical ML involving 5th digit with normal osseous elements.

Keywords: Macrodystrophia lipomatosa, 5th digit, fibrolipomatous hamartoma, localized proteus syndrome

1. Introduction

Macrodystrophia lipomatosa (ML) results from progressive overgrowth of all the mesenchymal elements, with a disproportionate increase in fibro-adipose tissue. This rare congenital abnormality occurs most frequently in the distribution of the median nerve in the upper, and medial plantar nerve in the lower, extremity.In ML, besides fat deposition in nerve sheaths, subcutaneous and muscle compartment, there is involvement of periosteum also leading to the bony changes, such as hypertrophy, exostosis, ankylosis of interphalangeal joints and fatty invasion of the medullary cavity. We report a case of atypical ML in distribution of ulnar nerve and involving 5th digit with no bony changes.

2. Case report

A twelve year old girl presented with history of progressive enlargement of little finger of right hand since birth. The growth of digit plateau at around the age of eight and has been stationary since then. There was slight restriction of movement of finger and reduced power of grip.(Fig 1-2) There was no history of swelling in any other part of body, and no family history. Overlying skin was normal, and there was no bruit. There was no sensory deficit or parasthesia over the digit. Plain radiograph of hand showed increased soft tissue shadow with normal bone and joints. MRI of hand showed diffuse enlargement of 5th digit and ulnar aspect of palm owing to enlarged soft tissue showing a striated pattern with fatty infiltration. The soft tissue displayed hyperintense signals on T2- and T1-weighted imaging (WI) with a few linear hypointense areas alternating with hyperintense areas on both T1- and T2- weighted images . On fat-suppressed short-tau inversion recovery (STIR) images, hypointensity was seen within the lesion suggestive of fatty components forming the bulk of the soft-tissue swelling.(Fig 3-4) There was infiltration of interosseous muscle by fatty tissue and ulnar digital nerves could not be visualized separately.(Fig 5) Patient underwent debulking of digit and ulnar aspect of palm. Microscopic examination of the specimen revealed fat deposits, ligamentous, and neural tissue elements in normal structure. The reports were consistent with diagnosis of Macrodystrophia lipomatosa of 5th digit.

Fig 1: Clinical image showing enlargement of 5th digit. Dorsum view



Fig 2: Clinical image showing enlarged 5th digit and ulnar aspect of palm. Volar view



Fig 3: T1.weighted coronal MR image. Prominent Subcutaneous fat along the right little finger and palmar area.



Fig 4: T1.weighted axial MR image showing infiltration of palm by fatty tissue.

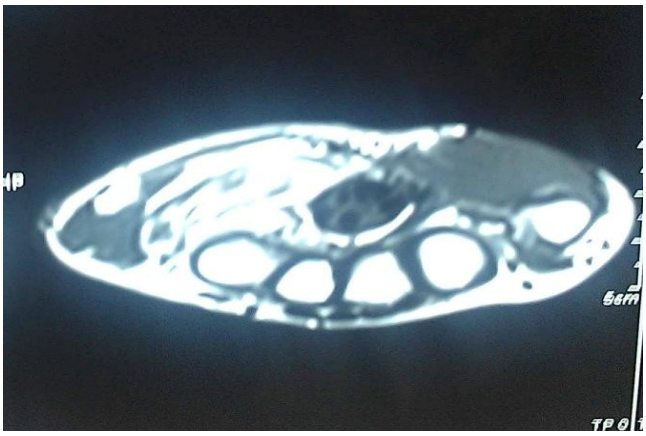


Fig 5: T2.weighted coronal MR image. Showin fatty infiltration of 5th digit.



3. Discussion

Four types of lipomatous masses associated with peripheral nerves have been identified: soft-tissue lipoma, intraneural lipoma, macrodystrophia lipomatosa, and lipofibromatous hamartoma[1]. Soft-tissue lipomas are true benign neoplasms originating from adipose cells, which can cause peripheral nerve compression extrinsically[2]. Intraneural lipomas arise from adipose cells within the peripheral nerve. These benign tumors are encapsulated, clearly defined, and easily excised[3] . Lipofibromatous hamartoma, is a rare condition characterized by diffuse enlargement of a nerve trunk, caused by overgrowth and fibrofatty tissue proliferation within the epineurium, perineurium, and endoneurium without overgrowth of the surrounding tissues. The individual nerve fibers are normal by microscopy[4].

Macrodystrophia lipomatosa (ML) is a developmental anomaly which has also been called as partial acromegaly, macrosomy, elephantiasis, megalodactly, dactylomegaly, macrodactly and limited gigantism. In 1925 this condition was first described by Feriz, who used the term macrodystrophia lipomatosa to refer to localised gigantism of the lower limb. Golding extended the term, in 1960, to include involvement of the upper limb[5].

The precise aetiology is not known; however, there are many hypotheses suggesting that it is due to fibrofatty tumours impinging on nerves supplying the enlarged digit, or due to alteration of somatic cells during limb bud development, disturbed foetal circulation and disturbance of growth factor in utero. Some authors suggest that ML is a localized form of Proteus syndrome . Recent genetic studies have indicated involvement of the PTEN (phosphatase and tensin homolog deleted on chromosome 10) gene, which is primarily a tumor suppressor gene and substantially influences cell/tissue growth[6]. PTEN mutations and PTEN nullizygosity have already been reported in Proteus and Proteus-like syndromes, which—in addition to hemihypertrophy, lower limb asymmetry, and arteriovenous malformations also presents with lipomatosis[7-8] .

The characteristic feature of the condition is that the anatomical location of the abnormality corresponds closely with the zone of innervation by the sclerotome. Involvement of the 2nd and 3rd digits of the hands and feet is common, corresponding to the median and the medial plantar nerve supply in the upper and lower limbs respectively; but 5th digit involvement is rare. Premature closure of epiphyseal ossification centers of phalanxes and metatarses, syndactly, polydactly, clinodactly, brachydactly and symphalangism may accompany ML. Involvement of lower extremity is encountered more frequently than involvement of upper extremity.

There is usually a long history of a painless mass since childhood. Patient may be aware of a painless mass for years. Patients may have signs and symptoms of nerve compression in the distribution of the affected nerve characterized by paraesthesia, motor deficit and pain. Patient may present with abnormally large finger(s) (macrodactyly). Bony abnormality may also occur.

Since overgrowth mainly involves the volar aspect, it can produce dorsal deviation of affected parts. Malalignment of joint and repeated trauma can cause secondary osteoarthritis and compression of neurovascular structures[9]. Osteophyte overgrowth may also cause compression of adjacent nerves and vessels, most commonly seen is carpal tunnel syndrome . Electroneurography and nerve conduction velocity tests may reveal slowed distal motor and sensory conduction, local (segmental) conduction block or slowing of the peripheral nerves at the entrapment sites[10].

Two subtypes of congenital macrodystrophia lipomatosa exist. These are static and progressive cases. In static cases the growth of the enlarged digit(s) is at the same rate as the other digits. In progressive cases the growth of the enlarged digit(s) is more rapid than the rest of the extremity. The progressive form is the less common. Involvement of the metacarpal and metatarsal bones is more likely in the progressive group; however in both groups the changes are most pronounced at the distal end of the digit (s)[11].

Differential diagnosis of ML includes neurofibromatosis type 1 (plexiform neurofibroma), fibrolipomatous hamartoma (FLH), lymphangiomatosis, hemangiomatosis and Klipe-Trenaunay-Weber syndrome, Mafucci syndrome, Ollier disease as well as Proteus syndrome. There is no familial predominance or neurocutaneous involvement in ML as seen in neurofibromatosis. Besides involvement in neurofibromatosis is usually bilateral and involvement of distal phalanx is not severe. Significant skin pathologies like capillary hemangiomas and varicose veins are seen Klippel-Trenaunay-Weber (KTW) syndrome. Although Mafucci Syndrome resembles KTW syndrome, soft tissue varicose veins are not seen in Mafucci syndrome. Diffuse swelling and pitting edema are present in cases of lymphangiomatosis. Dermatologic findings as well as palmar and plantar cerebroid thickening present in Proteus syndrome is not seen in ML.

Fibrolipomatous hamartoma (FLH) is another heterogeneous group of lesions which produces digital overgrowth

and can be confused with ML. It usually presents as an isolated nerve lesion and associated with intramuscular fat deposition . But in ML, besides fat deposition in nerve sheaths, subcutaneous and muscle compartment, there is involvement of periosteum also leading to the bony changes, such as hypertrophy, exostosis, ankylosis of interphalangeal joints and fatty invasion of the medullary cavity.

In conventional X-Ray images macrodactyly and soft tissue overgrowth are visible and are most marked along the volar aspect of the digit and at its distal end. This overgrowth can produce dorsal deviation of affected parts[12]. The phalanges are elongated, broad, and the distal ends are splayed and can have a "mushroom" shape[5]. Slanting of the articular surfaces can occur and this leads to secondary degenerative joint disease, manifesting in subchondral cyst and osteophyte formation. Sonography reveals alternating hyperechoic (fat) and hypoechoic (nerve fascicles) bands in a diffusely enlarged nerve, thus creating a cable- like appearance[13].

MRI demonstrates an excess of fibro-fatty tissue around the affected digits. This will therefore have the same signal characteristics as fat on MRI; i.e. high signal on T1 and T2 weighted sequences and low signal on hit suppression sequences (STIR). Fibrous strands within the fatty tissue will be demonstrated as low signal linear strands on TI weighted sequences. The fatty tissue may also be seen to infiltrate the adjacent muscles. Any bony abnormalities such as cortical thickening and secondary degenerative changes are also identified with MRI. Fibrous thickening of a nerve may also be seen[14].

In haemangiomatosis T2 weighted MR imaging shows increased signal from the serpiginous vascular channels within the haemangiomas[15]. In lymphangiomatosis, the lymphangiomas are hyperintense to muscle on T1 weighted images and hyperintense to fat on T2 weighted images[16].

In lipofibromatous hamartoma the nerve is abnormally enlarged, with the fascicles appearing as a coaxial cable in low-signal intensity on both T1- and T2-weighted axial images; the fascicles are separated from one another by uniformly dispersed fat, which appears on MRI as high-signal intensity on T1-weighted images and as low-signal intensity on fat-suppressed T2-weighted images. On coronal section, the nerve has a spaghetti or strands-of-hair appearance, again in low-signal intensity and embedded in fatty tissue[17].

Lipofibromatous hamartoma can be differentiated from other nerve lesions associated with focal or diffuse nerve enlargement, including intraneural lipoma, ganglion cyst, traumatic neuroma, plexiform neurofibroma, and hereditary hypertrophic interstitial neuritis of Dejerine-Sottas diseaseby their high-signal intensity on T2-weighted images. When fat is evenly distributed among nerve fascicles in conjunction with low-signal intensity on T2-weighted images, the diagnosis of lipofibromatous hamartoma can be made[18-19]. Fibrolipomatous hamartoma of the nerve may also be one of the features of macrodystrophia lipomatosa[20], which occurs in about 25% patients with fibrolipomatous hamartoma[21].

On histopathological examination there is increase in adipose tissue scattered in a fine lattice of fibrous tissue, which involves the bone marrow, periosteum, muscles, nerve sheaths, and subcutaneous tissues. The periosteum can be seen studded with small nodules of chondroblasts, osteoblasts, and osteoclasts. These nodules become larger and more numerous toward the distal ends of the phalanges making them elongated, broadened, and splayed at the distal ends often called mushroom-like appearance. There may be hypertrophy and degeneration in bone structure and increase in the medullary bone density. Trabecular structure of bones is characteristically well-preserved[5].

Treatment is aimed at cosmetic improvement and preserving neurological function[22]. Surgical interference is indicated depending upon patient's symptoms, age, and extent and severity of the disease. For lesion involving median and plantar region , conservative approach has been advocated . In localized diseases involving single digits and causing mechanical obstruction can be managed with excision of ray which can lead to a cosmetic improvement and does not cause any functional or neurologic problems as well. Multiple debulking procedures, epiphysiodesis, and various osteotomies are indicated for more severe form of the disease. Epiphysiodesis is not an effective method when performed alone as it may lead to loss of function. Sometimes ML involving whole of limb can be challenging where attempts a limb salvage needs to be made[23]. However, midtarsal or midcarpal amputations[24], or even more drastic amputations such as the below-knee amputation have been reported as necessary procedures for successful management of progressive macrodystrophia lipomatosa[25]. Besides, the incidence of nerve injury following extensive debulking or lesion removal is approximately 30%-50%. Another upsetting fact is the high recurrence rate which to the tune of 33%-60% [26].

Treatment of lipofibromatous hamartoma is controversial because the lesion cannot be completely excised without sacrificing the involved nerve. However, the benign tumor can be significantly debulked to improve functional outcomes.

Whereas some authors have reported that excision of the involved nerve did not lead to apparent detriment to sensibility[27], others have reported neural deficit[28].Therefore, excision of the nerve is not recommended. Extensive intraneural dissection may lead to neurologic deficit caused by ischemic complications[29].

4. Conclusion

ML is progressive enlargement of the fibrofatty tissue involving all the layers of soft tissue and even bone more commonly leading to localized gigantism. Diagnosis is accomplished on the basis of clinical and radiological evaluation MRI is most useful imaging modality in differentiating it from lipofibromatous hamartoma and other swellings of digits. Once diagnosed all attempt must be made in digit salvage, restoration of function and aesthetic value of digit.

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