ABSTRACT

Introduction: Macrodystrophia lipomatosa (MDL) is a non-hereditary congenital developmental anomaly characterized by localized gigantism due to overgrowth of all mesenchymal elements. Case Report: The authors describe a case of macrodystrophia lipomatosa (MDL) with involvement of the median nerve in a 10-year-old girl and review the pathologic, radiographic, ultrasonographic and MR imaging characteristics. Conclusion: Ultrasonography and MR imaging are pathognomonic for MDL and reliably distinguish it from other forms of localized congenital gigantism. However, since ultrasonography has the advantages of much lower cost, portability and easy accessibility, we propose its use as the imaging modality of choice in the initial diagnostic approach of congenital localized gigantism.

Keywords: Macrodystrophia lipomatosa, Localized gigantism, Macrodactyly, Ultrasonography

INTRODUCTION

Macrodystrophia lipomatosa (MDL) is a non-hereditary congenital developmental anomaly characterized by localized gigantism due to overgrowth of all mesenchymal elements [1]. It is characterized by an irregular increase of fibroadipose tissue in a neural distribution. The term MDL was initially coined by Feriz in 1925 to describe cases of lower-extremity gigantism that were associated with massive overgrowth of the accompanying adipose tissue [2]. Although MDL was originally described as primarily involving the peripheral parts of the involved
extremities, it is now known that proximal parts of the extremities can also be affected.

We describe a case of MDL with involvement of the median nerve in a 10-year-old girl who was operated in another institution due to carpal tunnel syndrome and describe the pathologic and imaging characteristics of this condition.

CASE REPORT

A 10-year-old girl was referred to us for an orthopedic and oncologic consultation by her pediatrician due to macrodactyly of the right ring finger. According to mother, the child's right ring finger started to grow at the age of six months. Over the last year along with the macrodactyly, she had noticed soft swelling of the ipsilateral volar aspect of the palm. Due to tingling in the distribution of the right median nerve, she was operated in another institution for carpal tunnel syndrome. Although she did not appreciate substantial improvement of her symptoms, she sought medical attention to us mainly for cosmetic reasons, since she was embarrassed from the macrodactyly to the point of avoiding school. Prior medical and family history was unremarkable. The initial physical examination confirmed the presence of macrodactyly (Figure 1) that was associated with the presence of soft, fibrolipomatous tissue in the outer half of the right palm. The remaining examination showed no cutaneous stigmata or organomegaly. Moreover, there were no bruits or edema in the involved extremity. Review of the slides from the tissue removed at surgery for carpal tunnel syndrome demonstrated excessive thickening of the perineurium of involved nerves, a characteristic finding of MDL (Figure 2). Plain radiographs showed soft tissue thickening and broadening of the second phalanx of the ring finger (Figure 3). After the initial examination, sonography was obtained that showed alternating hyperechoic and hypoechoic bands in a diffusely enlarged median nerve (Figure 4A and 4B).

Finally, MR imaging demonstrated longitudinally oriented cylindrical areas of low to intermediate signal intensity surrounded by adipose tissue in a diffusely thickened right median nerve, along with fat deposits in the muscles of the volar aspect of the right palm (Figure 5).

The patient remains mildly symptomatic, but after counseling and intense psychological support, she returned to school.

DISCUSSION

MDL is commonly associated with fibrolipomatous infiltration of peripheral nerves, with the median and plantar nerves being the most frequently affected [1-3]. In these cases, we talk about fibrolipomatous hamartoma, i.e., an anomalous growth of fibroadipose
tissue of the nerve sheath. Although fibrolipomatosis of the nerve can occur in isolation, i.e., without associated localized gigantism, in up to two thirds of the cases there is associated macrodactyly, and these cases are indistinguishable from MDL. Despite that, in MDL one entire extremity may be affected and abnormal fat deposits are not limited within the nerve sheaths but also involve the subcutaneous tissues, periosteum, and muscles. Some suggest that rather than MDL, the most appropriate term to describe lesions with associated localized gigantism is fibrolipomatous hamartoma of the nerve with macrodactyly [3]. Although overgrowth of all mesodermal elements of the digits is the dominant finding in MDL, associated clinical findings such as polydactyly can occur [1].

In most reported cases to date, the lesions are present at birth or develop within the first weeks of life. The lesions are unilateral and associated with uneven and progressive overgrowth that is more rapid compared to the rest of the limb. Cases with bilateral involvement have been described, but are extremely rare [4].

Radiographs reveal soft-tissue and osseous overgrowth often with elongated, broadened, and splayed phalanges [5]. The imaging appearance, particularly with sonography and MR imaging of advanced lipomatosis of a nerve is usually distinctively characteristic and reflects the underlying disease. Sonography reveals alternating hyperechoic (fat) and hypoechoic (nerve fascicles) bands in a diffusely enlarged nerve, thus creating a cable-like appearance [6, 7]. MR images are similar, with longitudinally oriented cylindrical areas of low to intermediate signal intensity (nerve fascicles) surrounded by adipose tissue in a diffusely thickened nerve [8]. On MR images, increased fat content of the digits is also apparent in patients with macrodactyly. MR imaging is considered
CONCLUSION

In conclusion, we describe a case of progressive macrodactyly due to MDL and present its pathologic and imaging findings. Even though history and physical examination are usually diagnostic, both ultrasonography and MR imaging can reliably distinguish MDL from other forms of localized congenital gigantism. However, since ultrasonography has the advantages of much lower cost, portability, and easy accessibility, we propose its use as the imaging modality of choice in the initial diagnostic approach of congenital localized gigantism.

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Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

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REFERENCES


