IMAGES IN NEONATAL MEDICINE

Macrodystrophia lipomatosa in a healthy newborn

A 15-day-old boy presented with an isolated enlargement of his second left toe. Physical examination showed no oedema, no bruit, no marks of amniotic constriction bands, or changes in the skin or the toenail. Afterwards, the toe exhibited a disproportionate growth with a progressive lateral deviation and dorsal flexion (figure 1). Radiography showed an increase in size that involved the bone and the soft tissues (figure 2).

Feriz first used the term macrodystrofia lipomatosa (MDL) to describe an unusual form of gigantism of the lower extremity.[1] The most frequent clinical pattern is the unilateral involvement of a lower limb, usually the second or the third toes. It preferentially affects the distal and volar aspects of the fingers. It may be associated with other anomalies as polydactyly or syndactyly[2 3] or it can be an isolated finding in a patient otherwise healthy as in this case. It has recently been linked to PIK3CA mosaicism.[4]

The diagnosis can be confirmed by radiological tests and histopathology.[5]

Although MDL is a benign condition, progressive deformities can cause physical impairment and interfere with daily activities.[5] MDL is also related to premature degenerative joint changes and to neurovascular compression. Surgical intervention is the mainstay of the treatment. As progressive overgrowth and deformity was observed, amputation of the second ray was performed at 17 months of age. The patient actually is 3 years old and has no functional limitations or neurological sequelae.

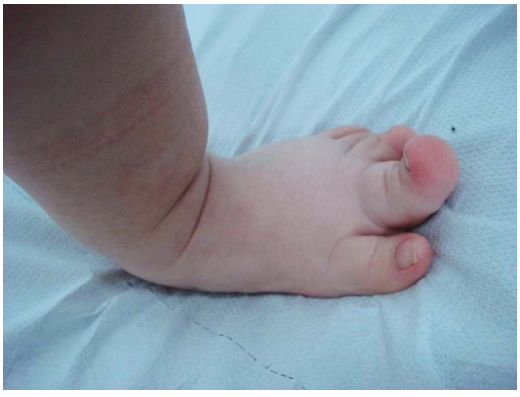

Figure 1 Disproportionate enlargement with dorsal flexion of the distal phalanx (12 months old).

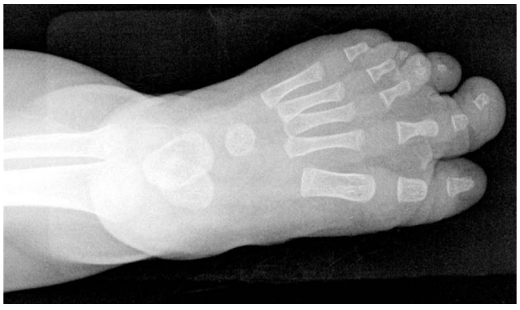

Figure 2 Radiograph, showing involvement of the bones and the soft tissues (4 months old).

Follow-up is mandatory to detect early recurrences or additional signs that could lead to the diagnosis of a syndromic macrodactyly.

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