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Behind neurofibromatosis type 1: an unusual finding

Detrás de la neurofibromatosis tipo 1: un hallazgo inusual

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We present the case of a girl aged 9 months referred for evaluation of café-au-lait spots present from birth with progressive increase in their number.

She exhibited normal psychomotor development at the time of evaluation, meeting Sheridan's norms for age, with good weight gain and linear growth. There was no reported history of similar spots in family members.

The physical examination revealed the presence of more than 10 café-au-lait spots more than 5 mm in diameter. There were no ephelides. She had slightly raised, yellowish lesions measuring less than 1 cm on the scalp and back and asymmetry of the left lower extremity with pronounced bowing (Fig. 1). The plain radiograph evinced cortical thickening and narrowing of the medullary canal thinning, suggestive of tibial dysplasia with congenital pseudarthrosis (Fig. 2).

Genetic testing detected a heterozygous pathogenic variant, c.7486C > T p.(Arg2496Ter), in the *NF1* gene, with an autosomal dominant pattern of inheritance.

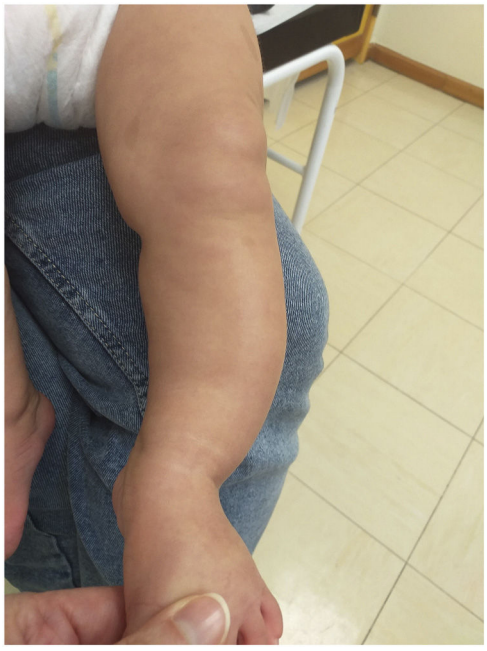

Figure 1 Left lower extremity asymmetry with pronounced bowing.

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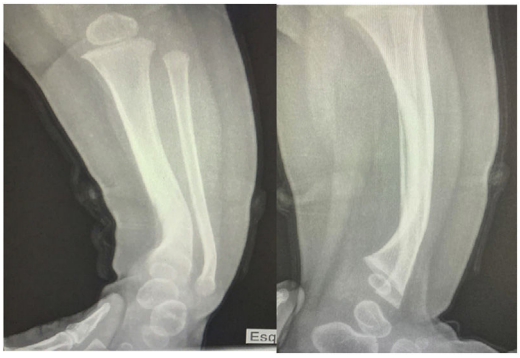

Figure 2 Cortical thickening and narrowing of the medullary canal suggestive of tibial dysplasia with congenital pseudarthrosis.

Despite not having parents with known disease, she met the criteria for diagnosis of neurofibromatosis (NF).[1,2]

We ought to highlight the rarity of a finding such as tibial dysplasia and pseudarthrosis, which only occurs in about 5% of children with a diagnosis of NF type 1.[2] These children are at higher risk of bone fracture.[2]

It is important to guarantee a multidisciplinary follow-up and access to appropriate treatment for these patients.[3]

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