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A CASE OF NEUROFIBROMATOSIS TYPE 1 DIAGNOSED AT A PEDIATRIC RHEUMATOLOGY SERVICE

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BACKGROUND

Neurofibromatosis type 1 (NF-1) is an autosomal dominant genetic disorder with a prevalence ranging from 1:2,000 to 1:7,800 live births. Occurring more frequently in childhood, the clinical criteria proposed by the National Institute of Health (NIH) Consensus Development Conference are used for diagnosis, considering the presence of two or more of the following characteristics: six or more café au lait spots (CALS) measuring > 5 mm in prepubertal children or > 15 mm in post-pubertal individuals; two or more neurofibromas; freckles in the axillary or inguinal regions; optic glioma; two or more Lisch nodules; bone lesions (long bone pseudoarthrosis or sphenoid wing dysplasia); an immediate family member with NF-1.

CASE REPORT

A 10-year-old prepubertal girl complained of arthralgia in the hands, feet and knees, moderate intensity, for approximately one year, with no signs of arthritis or improvement using simple analgesics. For two years, she had presented diffuse cutaneous lesions of brownish coloration without receiving any medical attention or diagnosis. The patient referred to the pediatric rheumatology service on suspicion of idiopathic juvenile arthritis (IJA); however, upon evaluation she did not meet these clinical criteria. On physical examination, more than 15 diffuse CALS, measuring > 5 mm, were observed, as well as a nonpainful bulge in the right scapular region, of fibroelastic consistency, measuring approximately 3 × 3 cm. The suspicion of lipoma or neurofibroma was not confirmed by chest tomography, on which the bulge was not evidenced. Physical and radiographic examinations of the spine indicated scoliosis. Due to the suspicion of neurofibromatosis, echocardiography and magnetic resonance imaging were solicited, yet no abnormalities were detected. Fundoscopy revealed multiple Lisch nodules. Neuropsychomotor development and weight and height gain were deemed suitable for age, with no similar cases reported in relatives.

CONCLUSION

Despite the onset of presentation occurring two years prior, the patient was diagnosed with NF-1 only following referral to a tertiary rheumatology service, which calls attention to the need for global evaluations patients with musculoskeletal complaints. The early suspicion of NF-1 is essential when following patients at specialized centers, considering that the risk of developing complications and/or neoplasms. Although relatively few lesions evolve to malignancy, NF-1 may be associated with higher mortality if not diagnosed early.