

CASES FOR DIAGNOSIS

Bilateral Symmetrical Nail Hypoplasia of the Hands

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Clinical History

We present the case of a 15-month-old boy who was brought by his mother for lesions affecting the nails of the first 3 fingers of both hands, and that had been present since birth. The other fingernails and the toenails showed no abnormalities. The patient had started walking at 1 year, with abnormalities of gait and frequent falls. In addition, the child had started speaking late, at 12 months of age, saying only individual words at the age of 15 months. The mother stated that her other daughter and several members of the paternal family had similar abnormalities of the nails, also associated with gait disturbances (Figure 1).

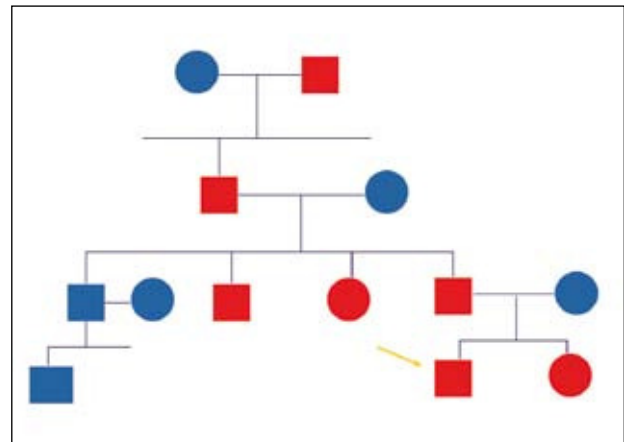


Figure 1.

Physical Examination

Physical examination showed symmetrical abnormalities of the nails of the first 3 fingers of both hands, most marked on the thumbnail, with hyponychia, thinning, and onychorrhexis, associated with triangular lunulae (Figure 2).



Figure 2.

Additional Tests

Renal ultrasound and blood and urine tests were performed, showing no abnormalities. Ophthalmologic examination also revealed no pathology and, finally, x-rays of the vertebral column, limbs, iliac crests, and hips showed the presence of posterior iliac horns (Figure 3).



Figure 3.

What Was the Diagnosis?

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Diagnosis

Nail-patella syndrome, hereditary osteo-onychodysplasia, or Fong syndrome.

Discussion

The nail-patella syndrome is a rare disorder of autosomal dominant inheritance with variable penetrance and expression. It is characterized by 4 major signs: dysplastic nails with triangular lunulae, hypoplastic or absent patella, posterior iliac horns, and hypoplasia of the head of the radius. Not all these signs are present in all cases. Other possible alterations include ocular and renal disorders, arterial aneurysms, psychomotor delay, and convulsive crises. Nail changes are observed in 98% of cases. The dysplastic nails are present at birth.¹ Typically, the nails of the first 3 fingers of the hands are more intensely affected, with alterations such as hypoplasia, absence, concave nails, and the presence of longitudinal ridges. The presence of triangular lunulae is a characteristic finding. In addition, there may be hypoplasia of the distal phalanges.² Congenital hyponychia and anonychia are rare malformations that can form part of syndromes such as the nail-patella syndrome, ectodermal dysplasias, or brachydactyly, or they can occur as isolated findings. These abnormalities are frequently associated with underlying skeletal disorders.³ The notable bone findings are posterior iliac horns, usually bilateral and present in 80% of cases,⁴ and dysplasia of the elbow and patella (in more than 90% of cases), giving rise to subluxations and early arthritis. Hypoplasia of the scapulae may also be present.² It should be noted that the patella does not ossify completely until late childhood, and, depending on the age of the patient, ultrasound should be used in order to evaluate the structure. There is associated nephropathy in 25% to 50% of cases; this is the major complication due to the possibility of renal failure, which occurs in up to 10% of cases. The renal

disease presents mainly as asymptomatic proteinuria or glomerulonephritis, with hematuria, albuminuria, and hyaline casts in the sediment.^{1,5}

The eye changes that may be found include hyperpigmentation of the pupillary border of the iris, as well as cataracts, keratoconus, microphakia, microcornea, and glaucoma.^{2,6}

The disease is caused by an alteration of gene *LMX1b*, present on chromosome 9, and which is involved in the development of the limbs, kidneys, and eyes. The presence of the alteration of this gene is not related to the prognosis, and its detection is not useful in prenatal diagnosis. The involvement of other genes, responsible for the variable expression of the disease, has been suggested.¹

When this diagnosis is suspected, bone, renal, and ocular evaluations must be performed, together with periodic follow-up in order to reach the diagnosis and start early management of the complications that may arise due to the syndrome.

Conflicts of Interest

The authors declare no conflicts of interest.

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