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Case Report

Peripheral Spondyloarthritis in a Patient With Noonan's Syndrome☆

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ABSTRACT

Noonan's syndrome is an autosomal dominant genetic disorder with high phenotypic variability, characterized mainly by facial dysmorphism, congenital heart disease and short stature. We describe the case of a male patient diagnosed with Noonan's syndrome and peripheral spondyloarthritis, a previously undescribed association in the literature.

Presentación de espondiloartritis periférica en paciente con síndrome de Noonan

Palabras clave:
Síndrome de Noonan
Espondiloartritis
Síndrome genético
Artritis

RESUMEN

El síndrome de Noonan es un síndrome genético autosómico dominante que presenta una gran variabilidad fenotípica, caracterizado principalmente por dimorfismo facial, cardiopatía congénita y talla baja. Describimos el caso de un paciente de sexo masculino con síndrome de Noonan y espondiloartritis periférica, asociación no descrita en la literatura hasta el momento.

Introduction

Noonan's syndrome (NS) is an autosomal dominant genetic syndrome characterized by short stature, cardiac abnormalities, a short neck, chest deformity, a characteristic phenotype with hypertelorism and mental retardation.[1] Its incidence so far has been estimated at 1:1000 to 1:2500 live births.[2]

The clinical characteristics of patients with NS are a triangular face with a broad forehead, hypertelorism, epicanthus, ptosis, a depressed nasal bridge, micrognathia, small ears, short neck and cardiac abnormalities.[3,4] Among the musculoskeletal disorders, the most common chest deformities are pectus carinatum and/or pectus excavatum, observed in 70% of cases. In addition there is ulnar valgus clynobrachydactylia, scoliosis/kyphosis, radioulnar synostosis, hyperextensibility and dental malocclusion.[5]

Other clinical findings are usually bilateral cryptorchidism, which occurs in 60% of male patients, learning and language disorders, attention deficit and depression in 23% of cases.[6]

NS diagnosis is made with clinical findings, according to the criteria formulated by van der Burgt in 1997 and published in 2007, described in Table 1.[7] Treatment is based on the clinical manifestations.[8]

We report the case of a young male patient with a clinical diagnosis of NS, exhibiting peripheral spondyloarthritis. The relevance of this case is the presentation of a genetic disease with spondyloarthritis, entities that generally do not coexist, allowing us to do a literature review and report this as the first clinical association seen in our region.

Clinical Case

The patient is a 23-year-old male with a family history and diagnosis of NS since the age of 2, manifested by cardiac

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Table 1
Van der Burgt Criteria for the Clinical Diagnosis of Noonan's Syndrome.

Characteristics	Major criteria	Minor criteria	Patient criteria
Facial	Typical face	Suggestive face	Typical face
Cardiac	Pulmonary valve stenosis Hypertrophic cardiomyopathy and/or ECG changes	Other cardiac abnormalities	Intraventricular communication which was treated surgically At the time, no cardiac abnormalities
Height	<3rd percentile	<10th percentile	<3rd percentile
Thoracic	Pectus carinatum and/or pectus excavatum	Width	Pectus carinatum
Family history	First-degree relative diagnosed with Noonan's syndrome	First degree relative with characteristics suggestive of Noonan's syndrome	First degree relative with characteristics suggestive of Noonan's syndrome
Other	Had 3: Mental retardation Cryptorchidism Lymphatic dysplasia	Had one: Mental retardation Cryptorchidism Lymphatic dysplasia	Mental retardation

Diagnosis of Noonan's syndrome:
-Two major criteria or one major + 2 minor criteria or 3 minor criteria.
-Typical face with hypertelorism, antimongoloid deviation of palpebral fissures, epicanthus, low-set and rotated ears.

abnormalities (ventricular septal defect diagnosed at 6 months, managed with digoxin, furosemide, antibiotic prophylaxis and surgery until resolved), chest deformity (pectus carinatum), cryptorchidism, bilateral inguinal hernia and surgically corrected phimosis, as well as short stature, a characteristic phenotype with hypertelorism and learning disorders. In 2013 he began a 6 months evolution with ankle, metacarpophalangeal and carpal inflammatory pain, with morning stiffness, and was treated with NSAIDs occasionally with little response; so he was referred to the rheumatology department, with joint pain, bilateral ankle edema and conjunctival hyperemia (Fig. 1).

On physical examination, he had a triangular face with a broad forehead, antimongoloid deviation of palpebral fissures, low-set and rotated ears (Figs. 2 and 3) He presented stunted growth. His height was 1.52 cm (<3rd percentile), weight: 38.4 kg (<3rd percentile), and he presented pectus carinatum, arthritis of the wrist and fourth metacarpophalangeal of the right hand, second and third proximal interphalangeal joint and distally on the left hand, knee as well as the right knee and ankle, with left anserine bursitis and enthesitis of the left Achilles tendon with bilateral plantar fasciitis. The modified Schober test was reduced (2 cm), thoracic expansibility 4 cm, lateral lumbar flexion 9 cm. Occiput–wall distance was 0 cm, tragus–wall 11 cm and finger–floor distance of 37 cm. The patient had no skin, cardiac or genitourinary abnormalities.

Laboratory tests showed: Hb 13.7 mg/dl, Hct 40.8%, WBC: 11,600/mm^3; platelets: 456,000/mm^3; neutrophils 59%, lymphocytes: 21% CRP: 27 3 mg/L; GOT: 13 U/L; GPT: 24 U/L, GGT: 36 U/L, urea 20 mg/dl, creatinine 0.8 mg/dl, rheumatoid factor: negative, HLA-B27: positive.

X rays of the knees and hands were performed and showed decreased joint space and osteopenia. Magnetic resonance imaging of the lumbosacral spine and hip showed normal vertebral bodies, spinal canal and interapophyseal and sacroiliac joints, with edema of the sacrum. MRI showed edema of the ankles in the posterior region of the calcaneus, thickening and increased density in the insertion of the left Achilles tendon, partial rupture and associated peritendinitis and retrocalcaneal bursitis (Fig. 4). Bone densitometry revealed a lower bone mass than expected for the patients' age.

The ophthalmologic evaluation revealed mild episcleritis, bilateral normal visual acuity, and the patient was treated with ophthalmic corticosteroid 2 times/day.

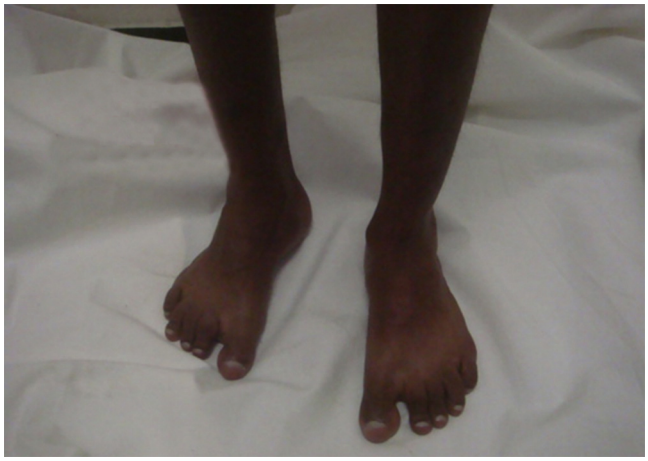

Fig. 1. Patient with Noonan's syndrome, with bilateral tibial edema.

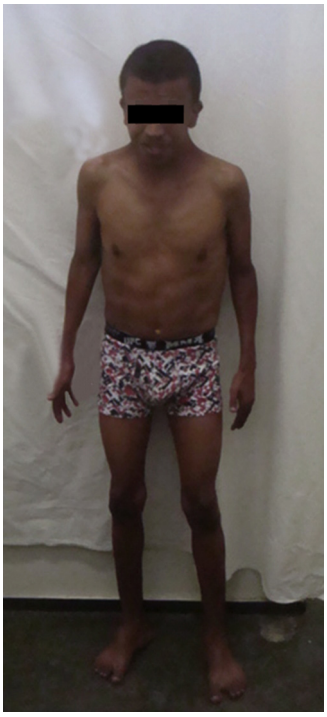

Fig. 2. Patient with Noonan's syndrome phenotype (short stature, short neck, chest deformity, and characteristic triangular face with broad forehead).