

Case Report

Pseudorheumatoid Dysplasia. A Rare Genetic Disorder Simulating Juvenile Idiopathic Arthritis[☆]

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ABSTRACT

We present the case of a teenage patient with progressive pseudorheumatoid dysplasia, an autosomal recessive disorder that may be initially misdiagnosed as juvenile idiopathic arthritis.

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Displasia progresiva seudorreumatoide. Desorden genético raro que simula artritis idiopática juvenil

RESUMEN

Adolescente con displasia progresiva seudorreumatoide, desorden autosómico recesivo que puede ser diagnosticado inicialmente como artritis idiopática juvenil.

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Introduction

Pseudorheumatoid progressive dysplasia (PRPD) is a hereditary, non-inflammatory skeletal disorder, autosomal, and recessive, associated to the WISP3. It is characterized by joint pain and effusion and progressively involves the hips, knees, wrists, and fingers. A reduction in joint mobility and multiple contractures lead to severe disability.

Case Report

A 13-year-old male patient presented pain of the right hip and left knee which had started when he was 13. Five months later he presented swelling of both knees and the interphalangeal joints of the right hand. The patient referred a history of difficulty walking since he was 3 years of age. Upon examination we found a normal teenager with abnormal gait, spinal scoliosis, and kyphosis of the thoracic spine. Mobility of the lumbar spine, coxofemoral joints, knees, ankles, and interphalangeal joints were severely limited. X-rays of the hands and feet (Fig. 1a and b), hips and knees (Fig. 2a and b) revealed diffuse osteopenia, tubular widening of the epiphysis and loss of joint space, without erosive lesions; X-rays of the hip and knees also showed degenerative changes, with widening of the femoral heads. Thoracic and lumbar vertebrae were flattened and had an irregular surface (Fig. 3a and b). C reactive protein and erythrocyte sedimentation rate were normal.

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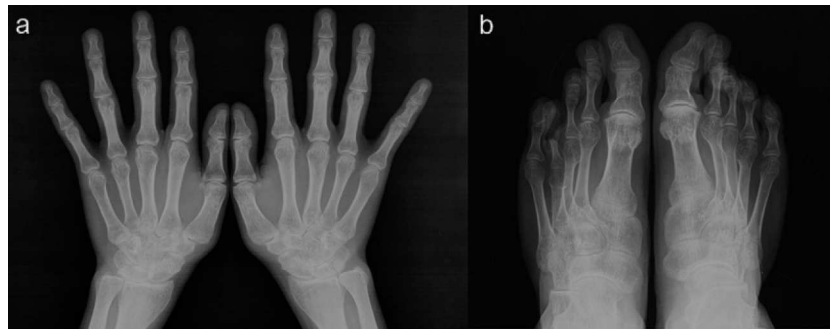


Fig. 1. (a and b) Hand and feet X-rays show osteopenia, tubular epiphyseal widening and loss of joint space, without erosions.

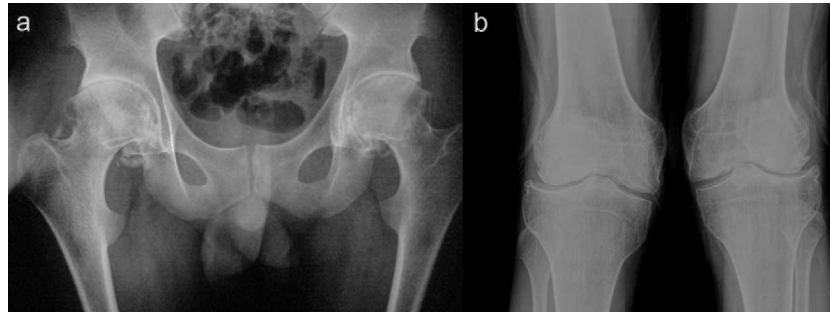


Fig. 2. (a and b) Coxofemoral and knee X-rays show severe degenerative changes.

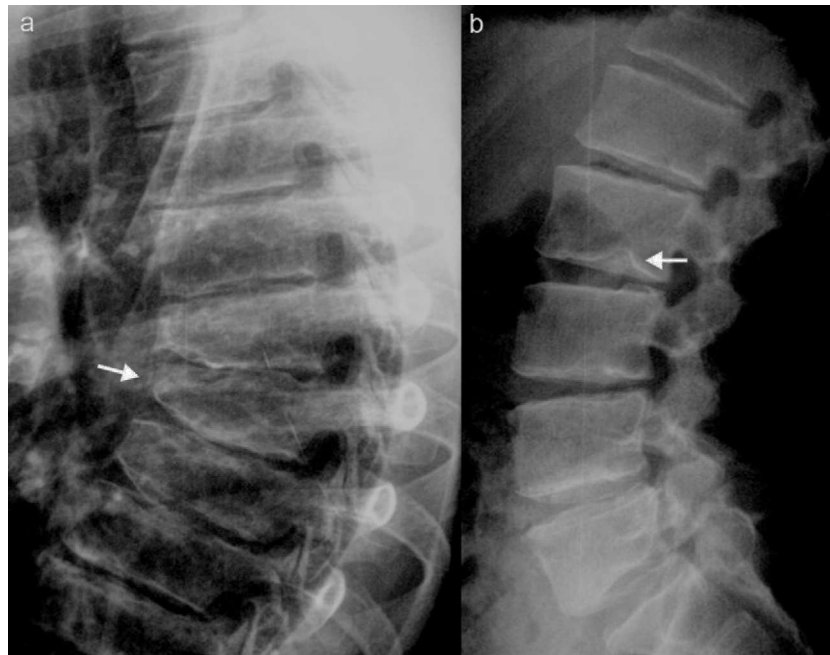


Fig. 3. (a and b) Flattened thoracic and lumbar vertebral bodies with irregularities of their surfaces (platyspondylyia).

Discussion

PRPD clinically simulates early stages of JIA^{1,2}; however, the evidence used to establish the diagnosis of bone dysplasia is non-inflammatory joint involvement and characteristic radiological findings, such as the presence of enlarged epiphyses, generalized osteoporosis, and platyspondylyia.³

This disease is caused by mutations in the WISP 3 gene, essential for normal growth and postnatal skeletal and joint cartilage homeostasis.⁴ Therefore, in contrast to most of the

skeletal dysplasias, prenatal skeletal growth and morphogenesis not altered in PRPD and affected individuals are asymptomatic during the first year of life. Subsequently, patients have progressive loss of joint cartilage and severe degenerative changes.^{4,5}

Ethical Responsibilities

Protection of Persons and Animals. No experiments were performed on humans or animals.

Data Confidentiality. No patient data appear in this article.

Right to Privacy and Informed Consent. No patient data appear in this article.

Conflicts of Interest

The authors have no conflicts of interest to make.

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