Spondyloepiphiseal dysplasia in a middle aged male

Displasia espondiloepifisiaria tarda en varón de edad media

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Introduction

Spondyloepiphiseal dysplasia represent a group of bone dysplasias that affect both axial and appendicular skeleton, which has a genetic origin and is characterized by epiphysial irregularities and deformity of vertebral bodies.

The case of a middle aged male is presented, with clinical and radiological data that suggest late spondyloepiphiseal dysplasia.

Case report

The patient is a 59 year old male with type 2 Diabetes Mellitus in treatment with oral antidiabetics, who came to the rheumatology clinic with bilateral inguinal, dorsal and lumbar pain of progressive intensity, both with mechanical characteristics, which the patient catalogued as very intense and which led to progressive functional limitation in walking, and by episodes of moderate to severe dyspnea, which was studied by the pneumologist, and were owed to a limited chest expansion. The pain had not been reduced when treated with non steroidal anti inflammatory drugs or minor opioids. Upon examination, the patient’s height is 146 cm, marked dorsal xyphosis, truncal obesity, important limitation for flexion and extension and rotation of both coxofemoral joints, as well as brachydactilia in both hands. Blood count and blood chemistry were normal. The conventional radiology studies of the dorsolumbar spine, pelvis and hands are shown in Figure 1, Figure 2, Figure 3 and Figure 4.

Discussion

The patient described presents data suggestive of a late spondyloepiphiseal dysplasia. This entity has an x chromosome linked inheritance, affecting therefore only males. Mutations have been seen on the SELD gene on chromosome Xp22.12-p 23.31. The diagnosis is performed between 5 and 10 years of age, presenting dwarfism with a short trunk; delay in growth of the spine with a mean final height
of 145 cm and disproportioned length between limbs and trunk. The coxofemoral joints is always affected as coxa vara with mild flattening of the epiphysis, leading to early degenerative changes and which may, in later stages, lead to disability. Axially, there are dysplastic changes in vertebrae, with platypondylia, leading to progressive xyphosis with an increase in the anteroposterior diameter of the thorax and which may ultimately lead to dyspnea. There is no mental retardation and familial forms have been described, in association with chondrocalcinosis, with the existence of a form late for of spondyloepiphyseal dysplasia existing, associated with early degenerative disease which may simulate juvenile rheumatoid arthritis.

References