



Images in clinical rheumatology

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 apendicular skeleton, which has a genetic origin and is characterized by epiphiseal 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 xiphosis, 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.



Figure 1. Lateral x ray of the thorax. Dorsal xiphosis, platyspondylia, irregularities of the vertebral body joint facets and a reduction in intervertebral discs.

Discussion

The patient described presents data suggestive of a late spondyloepiphiseal dysplasia. This entity has an x chromosome linked inheritance¹, affecting therefore only males. Mutations have been seen on the SELD gene on chromosome Xp22.12-p 23.31.¹ 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

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Figure 2. Lateral x ray of the lumbar spine. Platyspondylia as well as a humped appearance of the central and posterior portions of the vertebrae can be observed.



Figure 3. Anteroposterior x ray of the pelvis. There is shortening of both femoral necks, bilateral coxa vara and dysplasia of both femoral necks.



Figure 4. Posteroanterior x ray of both hands. There is brachydactyly of the second and third fingers with shortening of the second phalanges can be seen.

of 145 cm and disproportioned length between limbs and trunk.^{2,3} 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.²⁻⁴ Axially, there are dysplastic changes in vertebrae, with platyspondylia, leading to progressive kyphosis⁴⁻⁶ 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,^{7,8} with the existence of a form late for of spondyloepiphyseal dysplasia existing, associated with early degenerative disease which may simulate juvenile rheumatoid arthritis.⁹

References

1. Heuertz S, Nelen M, Wilkie AOM. The gene for spondyloepiphyseal dysplasia (SEDL) maps to Xp22 between DXS16 and DXS92. *Genomics*. 1993;18:100-4.
2. Bleasel J, Bisagni-Faure A. Type II procollagen gene (COL2A1) mutation in exon 11 associated with spondyloepiphyseal dysplasia, tall stature and precocious osteoarthritis. *J Rheumatol*. 1995;22:255-61.
3. Deere M, Blanton S, Scott C. Genetic heterogeneity in multiple epiphyseal dysplasia. *Am J Hum Genet*. 1995;56:698-704.
4. Hedden F. Spondyloepiphyseal dysplasia. *J Bone Joint Surg*. 1978;60B:295.
5. Gedeon AK, Colley A, Jamieson R. Identification of the gene (SEDL) causing X-linked spondyloepiphyseal dysplasia tarda. *Nat Genet*. 1999;22:400-4.
6. Sillence DO. Displasias esqueléticas congénitas. In: Behrman RE, Vaughan VC, editors. *Nelson. Tratado de Pediatría*. Philadelphia: Mc Graw-Hill; 1989. p. 1469-85.
7. Hamza M, Bardin T. Camptodactyly, polyepiphyseal and mixed crystal deposition disease. *J Rheumatol*. 1989;16:1153-8.
8. Sambrook PN, De Jager JP, Champion GD. Synovial complications of spondyloepiphyseal dysplasia of late onset. *Arthritis Rheum*. 1988;31:282-7.
9. Bal S, Kocyigit H, Turan Y. Spondyloepiphyseal dysplasia tarda; four cases from two families. *Rheumatol Int*. 2009;29:699-702.