

Progressive pseudorheumatoid chondrodysplasia of childhood

Shivanand G, Jain V, Lal H

ABSTRACT

Progressive pseudorheumatoid chondrodysplasia is an autosomal recessively inherited skeletal dysplasia, characterised by platyspondyly and progressive arthropathy. We report a 26-year-old woman who presented with a history of waddling gait, progressive restriction of movements of several joints and swelling of interphalangeal joints since her late childhood. She was diagnosed to have progressive pseudorheumatoid chondrodysplasia associated with synovial osteochondromatosis.

Keywords: arthropathy, pondoepiphyseal dysplasia, progressive pseudorheumatoid chondrodysplasia, skeletal dysplasia, synovial osteochondromatosis,

Singapore Med J 2007;48(5):e151–e153

INTRODUCTION

Progressive pseudorheumatoid chondrodysplasia was first described by Spranger et al.⁽¹⁾ Although rare, several additional cases have been reported since then, and it is now considered a distinct variety of osteochondrodysplasia with defined clinico-radiological features and genetic patterns.⁽²⁻⁷⁾ We describe a similar case, which also had synovial osteochondromatosis of glenohumeral and knee joints, in addition to progressive pseudorheumatoid chondrodysplasia. We believe that only one such case has been reported previously by Legius et al.⁽³⁾

CASE REPORT

A 26-year-old woman, born of consanguineous parents, presented with a history of waddling gait, progressive restriction of movements of several joints and swelling of interphalangeal joints since her late childhood. The patient had a family history of similar complaints affecting her younger brother, although her parents were apparently healthy. On clinical examination, the patient was short on stature. Her intelligence, vision, and hearing were normal. Movements of the hip, knee, ankle, wrist, elbow and shoulder joints were restricted on both sides. Her proximal and distal interphalangeal

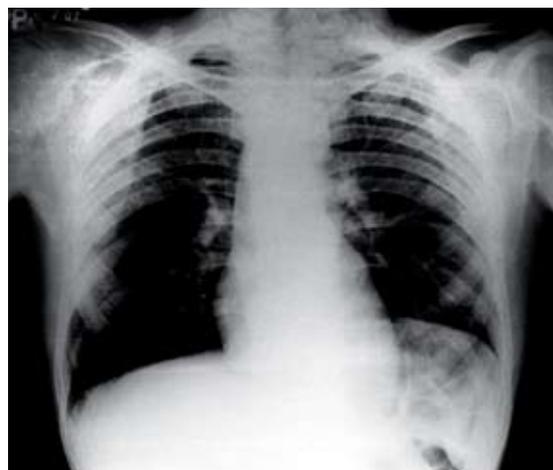


Fig. 1 Chest radiograph shows degenerative changes and synovial osteochondromatosis of both glenohumeral joints.



Fig. 2 Radiograph of the knee joints shows degenerative changes and synovial osteochondromatosis.

joints were swollen. Skeletal radiography revealed severe osteoarthritis of all the major joints, including the distal and proximal interphalangeal joints in both hands. In addition, there was synovial osteochondromatosis of both glenohumeral and knee joints (Figs. 1 & 2). Dorsal and lumbar spine revealed platyspondyly, multiple osteophytes and irregular endplates of most of the vertebrae (Fig. 3). Both femoral necks were short

Department of
Radiodiagnosis,
All India Institute of
Medical Sciences,
New Delhi 110029,
India

Shivanand G, MD
Assistant Professor

Jain V, MD
Senior Resident

Department of
Radiology,
Central Government
Hospital Services,
Kanpur,
Uttar Pradesh,
India

Lal H, MD
Specialist

Correspondence to:
Dr Shivanand
Gamanagatti
Tel : (91) 11 2658 8500
ext 4567
Fax: (91) 11 686 2663
Email: shiv223@
rediffmail.com

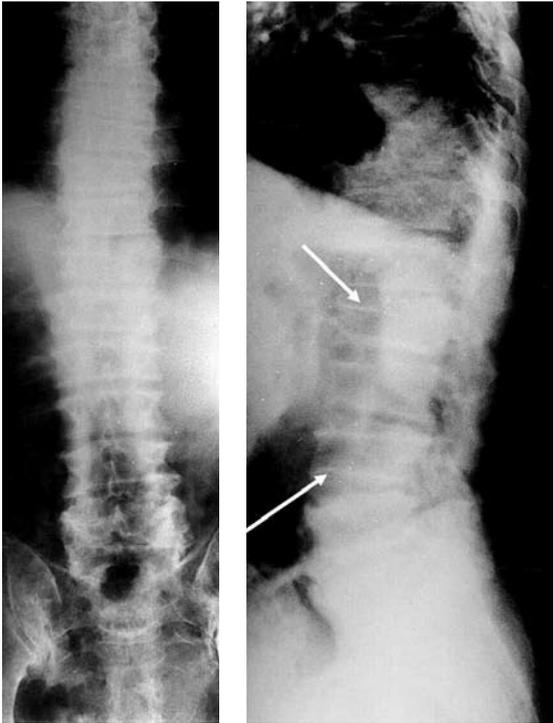


Fig. 3 (a) Anteroposterior and (b) lateral radiographs of the dorsolumbar spine show platyspondyly, spondylophytes involving multiple vertebrae and irregular endplates (arrows).



Fig. 4 Radiograph of the pelvis shows short femoral necks, irregular epiphyses and osteoarthritis of both hip joints.

and enlarged, with irregular epiphysis (Fig. 4). The skull was normal. Laboratory investigations revealed a normal erythrocyte sedimentation rate and anti-nuclear antibody titre. Based on clinical presentation and skeletal survey findings, the diagnosis of progressive pseudorheumatoid chondrodysplasia was made.

DISCUSSION

Progressive pseudorheumatoid arthropathy of childhood (PPAC) has other synonyms, like spondyloepiphyseal

dysplasia tarda with progressive arthropathy and progressive pseudorheumatoid dysplasia (PPD).⁽⁸⁾

Progressive pseudorheumatoid chondrodysplasia was first described by Spranger et al.⁽¹⁾ They suggested that it is a primary disorder of the joint cartilage. They demonstrated chondral abnormalities in an iliac crest biopsy with nests of clustered chondrocytes with pyknotic nuclei, irregular ground substance, and spiral bundles of thin fibre, mostly without striations. It is an autosomal recessive disorder in which patients exhibit platyspondyly and progressive arthropathy, resulting in short stature. Pain, swelling and stiffness of the joints are also characteristics, resembling the findings in rheumatoid arthritis, although without synovitis. There is no inflammation of the joints and it has been demonstrated that the disorder is actually due to a non-inflammatory chondropathy affecting mainly the articular cartilage. Early onset of cartilage loss is a very important element pathogenesis of the disease. The disorder usually presents between three and eight years of age with pain, kyphoscoliosis, easy fatigability, muscular weakness, progressive restriction of joint movement and swelling at several joints. Joint stiffness usually first affects the hips, and progressively involves the other joints, including the proximal and distal interphalangeal joints. In the illustrated case, the patient presented with a similar history, and clinico-radiological features suggestive of this rare disorder.

The incidence of this condition is unknown. For this autosomal recessive disorder, the gene has been localised recently to chromosome 6q22. Recently, it has been found that mutations in the CCN (CTGF, *cyr61/cef10*, *nov*) family member WISP3 are associated with the autosomal recessive skeletal disorder PPD.⁽⁹⁾ CCN gene family encodes cysteine-rich secreted proteins with roles in cell growth and differentiation, which is essential for normal postnatal skeletal growth and cartilage homeostasis.⁽⁹⁾ Due to its recessive inheritance, the disorder is more common in regions with a high incidence of consanguineous marriages, such as in South and Southeast Asia.⁽⁴⁾ The characteristic radiographical features of PPAC include narrow joint space with wide metaphyses and flat epiphyses, and enlarged femoral heads with irregular acetabular margins. Spinal abnormalities include platyspondyly with erosion of the endplates. This condition mimics juvenile rheumatoid arthritis but skeletal changes such as erosions, overgrowth of epiphyses, periostitis and osteopaenia are not seen in PPAC.

The usual variety of spondyloepiphyseal dysplasia tarda is an X-linked disorder. Premature osteoarthritis is a common complication in the large joints, particularly the hips. However, the peripheral skeleton is relatively unaffected, and the interphalangeal joints

are normal. Another condition that may be considered in the differential diagnosis is Stickler's syndrome. It is an autosomal dominant form of spondyloepiphyseal dysplasia with a variable expression. It also presents with multiple joint arthropathy. However, ophthalmic abnormalities, particularly progressive myopia, are usually associated with this disorder. Other disorders with platyspondyly, such as mucopolysaccharidosis, spondylometaphyseal dysplasia and rare forms of spondyloepimetaphyseal dysplasia, have relatively characteristic features, and are not confused with progressive pseudorheumatoid chondrodysplasia, because in these disorders, small joints of hand and larger joints like the knee are normal.

Spinal abnormalities mimic those of Scheuermann's disease. However, Scheuermann's disease presents at puberty whereas in PPAC, spinal abnormalities appear before the age of ten years. The pedicles are also shortened in PPAC. Magnetic resonance imaging findings of spinal abnormalities have been discussed by Mampaey et al, and include severe irregularities at the endplates of vertebral bodies with intravertebral and retromarginal herniations.⁽¹⁰⁾ In the illustrated case, spondyloepiphyseal chondrodysplasia was associated with progressive arthropathy and synovial osteochondromatosis affecting the shoulder and knee joints. To our knowledge, there is only one other case with such association, and that was reported by Legius et al in 1993.⁽³⁾ Primary or idiopathic synovial osteochondromatosis is an uncommon condition probably caused by synovial metaplasia and typically resulting in multiple calcified periarticular bodies. Synovial osteochondromatosis may also be seen secondary to other joint disorders such as osteoarthritis. We believe that the osteochondromatosis in our case is probably related secondary to osteoarthritis, and is therefore commoner

in large joints that have a large articular surface.

In conclusion, progressive pseudorheumatoid chondrodysplasia is a rare and less recognised entity, which may be associated with synovial osteochondromatosis due to progressive cartilaginous destruction. It may be confused with rheumatoid arthritis, both clinically and radiologically. In view of the difficulty in early clinical diagnosis of the disease, it is imperative that radiologists be familiar with radiographical features of the disease in order to avoid needless investigations and trials of medication.

REFERENCES

1. Spranger J, Albert C, Schilling F. A progressive connective tissue disease with features of juvenile rheumatoid arthritis and osteochondrodysplasia. *Eur J Pediatr* 1980; 133:186-9.
2. Spranger J, Albert C, Schilling F, Bartsocas C. Progressive pseudorheumatoid arthropathy of childhood (PPAC): a hereditary disorder simulating juvenile rheumatoid arthritis. *Am J Med Genet* 1983; 14:399-401.
3. Legius E, Mulier M, Van Damme B, Fryns JP. Progressive pseudorheumatoid arthritis of childhood (PPAC) and normal adult height. *Clin Genet* 1993; 44:152-5.
4. Rezai-Delui H, Mamoori G, Sadri-Mahvelati E, Noori NM. Progressive pseudorheumatoid chondrodysplasia: a report of nine cases in three families. *Skeletal Radiol* 1994; 23:411-9.
5. Al-Awadi SA, Farag TI, Naguib K, et al. Spondyloepiphyseal dysplasia tarda with progressive arthropathy. *J Med Genet* 1984; 21:193-6.
6. Wynne-Davies R, Hall C, Ansell BM. Spondylo-epiphysal dysplasia tarda with progressive arthropathy. A "new" disorder of autosomal recessive inheritance. *J Bone Joint Surg Br* 1982; 64:442-5.
7. Archik SG, Kamat RD. Progressive pseudorheumatoid chondrodysplasia simulating juvenile rheumatoid arthritis. *Indian J Pediatr* 1990; 57:785-8.
8. El-Shanti HE, Omari HZ, Qubain HI. Progressive pseudorheumatoid dysplasia: report of a family and review. *J Med Genet* 1997; 34:559-63.
9. Hurvitz JR, Suwairi WM, Van Hul W, et al. Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. *Nat Genet* 1999; 23:94-8.
10. Mampaey S, Vanhoenacker F, Boven K, Van Hul W, De Schepper A. Progressive pseudorheumatoid dysplasia. *Eur Radiol* 2000; 10:1832-5.