INTRODUCTION

Clinical manifestations of camptodactyly-arthropathy-coxa vara-pericarditis syndrome (CACP) include congenital or early-onset camptodactyly, childhood-onset noninflammatory arthropathy with synovial hyperplasia. Some patients have progressive coxa vara deformity and/or noninflammatory pericardial effusion. CACP is inherited as an autosomal recessive mode and the disease gene is assigned to a 1.9-cM interval on human chromosome 1q25-31. We describe a 10-year-old boy who has typical features of CACP without familial association.

CASE REPORT

A 10-year-old boy was admitted due to deformity of fingers and toes in August 2003. In July 1999, he was referred to our hospital because of joint swelling, pericardial effusion. Synovial fluid finding did not show inflammatory nature, and synovial biopsy revealed synovial hyperplasia without significant inflammation (Fig. 1). Ultrasound examination revealed hepatomegaly, but not splenomegaly. He had non-inflammatory arthropathy, pericardial effusion, and coxa vara deformity, and without familial aggregation.

Months prior to admission, camptodactyly of fingers and toes was developed and gradually progressed, and he was referred to our hospital again. Physical examination showed a blood pressure 100/60 mmHg, pulse rate 90/min, temperature 36.0°C, and normal respiration pattern. Swelling of both knees, ankles, elbows and wrists and deformity of fingers and toes were observed (Fig. 2). There was no evidence of fever, lymphadenopathy and skin rash. His family members did not have musculoskeletal abnormality. Laboratory findings revealed a white blood cell 5,500/µL; hemoglobin 10.9 g/dL; platelet 194,000/µL; erythrocyte sedimentation rate 9 mm/hr, C-reactive protein 0.1 mg/dL (reference range 0.1-0.8). Tests for rheumatoid factor, antinuclear antibody and HLA-B27 were all negative. Liver function test, serum creatinine, urinalysis were in normal ranges. Synovial fluid analysis from knee joints showed a white blood cell 5,500/µL, hemoglobin 10.9 g/dL; platelet 194,000/µL; erythrocyte sedimentation rate 9 mm/hr, C-reactive protein 0.1 mg/dL (reference range 0.1-0.8). Tests for rheumatoid factor, antinuclear antibody and HLA-B27 were all negative. Liver function test, serum creatinine, urinalysis were in normal ranges. Synovial fluid analysis from knee joints showed a white blood cell 5,500/µL, hemoglobin 10.9 g/dL; platelet 194,000/µL; erythrocyte sedimentation rate 9 mm/hr, C-reactive protein 0.1 mg/dL (reference range 0.1-0.8). Tests for rheumatoid factor, antinuclear antibody and HLA-B27 were all negative. Liver function test, serum creatinine, urinalysis were in normal ranges. Synovial fluid analysis from knee joints showed a white blood cell 5,500/µL, hemoglobin 10.9 g/dL; platelet 194,000/µL; erythrocyte sedimentation rate 9 mm/hr, C-reactive protein 0.1 mg/dL (reference range 0.1-0.8). Tests for rheumatoid factor, antinuclear antibody and HLA-B27 were all negative. Liver function test, serum creatinine, urinalysis were in normal ranges.
DISCUSSION

CACP is characterized by congenital or early-onset camptodactyly, childhood-onset noninflammatory arthropathy associated with synovial hyperplasia, progressive coxa vara deformity and noninflammatory pericardial or pleural effusion.

The definition of camptodactyly is a congenital or acquired nontraumatic flexion deformity of the proximal interphalangeal (PIP) joint of one or several fingers (3). Camptodactyly in CACP is usually bilateral and congenital, but in some cases, it develops in early childhood. The degree of contracture need not be equal in both and the deformity may progress or not improve (3). Camptodactyly may be present as an isolated entity or part of a spectrum of congenital anomalies. Camptodactyly may be present in congenital anomalies such as trisomy 13, oculo-dental-digital, oro-facial-digital, cerebro-hepato-renal, Catel Manzke, Pena-Shokeir I syndromes (3-5), and must be differentiated from a boutonniere deformity, Dupuytren's contracture, a trigger finger, congenital absence of the extensor mechanism (3). Nonoperative therapy is effective in managing camptodactyly. Splinting is a valuable tool in the initial management of the camptodactyly. Tenolysis and tenosynovectomy is beneficial in some patients (6, 7).

Arthropathy principally involves large joint such as elbows, hips, knees, and ankles. Synovial fluid analysis reveals non-inflammatory findings. Histopathologic analysis of synovial tissue reveals pronounced hyperplasia of synovium without evidence of inflammatory cell infiltration or vasculitis, while synovial hyperplasia in rheumatoid arthritis is associated with chronic inflammation. MRI finding of involved joints showed only prominence of cartilage with normal menisci and cruciate ligaments in one study (8), and rim-like enhancement
Camptodactyly, Arthropathy, Coxa vara, Pericarditis Syndrome

of the fluid filled bursae at T1 weighted image before and after contrast enhancement (9). Enhancement is related to the presence of inflammatory tissue, but the presence of rim-like enhancement means the noninflammatory features of CACP. The rim-like enhancement can distinguish between CACP and juvenile rheumatoid arthritis (JRA) based on the homogenous or multinucleated enhancement in JRA (10, 11). Therefore, MRI is regarded as a useful diagnostic tool which differentiates the CACP from other childhood connective tissue disease such as JRA (9).

The presence of coxa vara is noted in 50% of published CACP cases (1), and in one study, 90% of cases have coxa vara deformity (9). The long-term follow-up of CACP patients revealed the hip and spine involvement in some cases (12).

Noninflammatory pericarditis has been reported in up to 30% of CACP (1), and it may be mild and self-limited. But it may be necessary to perform a pericardiocentesis or pericardiectomy in life-threatening cases (2, 13-15).

CACP is a genetically homogenous condition despite clinical variability and differences in ethnic and geographic origins (12), and it has autosomal recessive mode of inheritance (2). A CACP locus is assigned to a 1.9-cM interval on human chromosome 1q25-31 by homozygosity mapping (1). Marcelino et al. identified mutations in a gene (CACP) encoding a secreted proteoglycan as the cause of CACP (16).

**Fig. 3.** The antero-posterior radiograph of pelvis shows coxa vara and short broad femoral neck.

**Fig. 4.** (A). The plain radiography of both hands shows flexion at the 5th PIP joint of both hands and questionable flexion at the 2nd PIP joint of left hand. (B) Radiography of both feet shows flexion at the left 2nd PIP joint.

**Fig. 5.** Sagittal T1 MR image of knee with gadolinium enhancement shows thin rim-like enhancement of the fluid filled bursae.

**Fig. 6.** Echocardiogram shows moderate amount of pericardial effusion posterior to left ventricle at apical 4 chamber view.
Although some cases of CACP were reported in Caucasian, Egyptian, Saudi Arabian, but there has been no report in Korea. The reason why CACP is rare in Korea may be due to rarity of consanguineous marriage, which increases the incidence of autosomal recessive disease. In this case, sporadic gene mutation might be responsible for the disease, because his family has no arthropathy or joint deformity. Whenever we see juvenile patients with noninflammatory arthropathy, congenital musculoskeletal disease such as CACP should be considered.

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