

Camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome

Mehmet Taşar¹, Zeynep Eyileten¹, Ferit Kasımcı¹, Tayfun Uçar², Tanıl Kendirli³, Adnan Uysalel¹

¹Department of Cardiovascular Surgery, Divisions of ²Pediatric Cardiology, and ³Pediatric Intensive Care, Department of Pediatrics, Ankara University Faculty of Medicine, Ankara, Turkey. E-mail: zeyileten@gmail.com

Received: 24 February 2014, Revised: 22 October 2014, Accepted: 10 November 2014

SUMMARY: Taşar M, Eyileten Z, Kasımcı F, Uçar T, Kendirli T, Uysalel A. Camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome. Turk J Pediatr 2014; 56: 684-686.

Camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome is a rare autosomal recessive congenital disorder that includes childhood-onset camptodactyly, synovial hyperplasia-related arthropathy, progressive coxa vara deformity and noninflammatory pericarditis. A seven-year-old male patient had a diagnosis of CACP. He had pericardial effusion and underwent surgical tube drainage. CACP syndrome is seen very rarely, and differential diagnosis is very important. CACP is usually treated medically, but surgery may sometimes be preferred, as in our patient.

Key words: pericarditis, familial arthropathy, camptodactyly.

Camptodactyly-arthropathy-coxa vara-pericarditis (CACP) syndrome is an autosomal recessive congenital disorder that is seen very rarely. This syndrome is associated with a mutation of the proteoglycan 4 (PRG 4) gene. It is characterized by early-onset camptodactyly and synovial hyperplasia with noninflammatory arthropathy, progressive coxa vara deformity and noninflammatory pericardial or pleural effusion. CACP syndrome mimics the most common rheumatic disorders, so it is usually labeled as juvenile idiopathic arthritis¹.

All of the components of this syndrome may not be detected on physical examination, so radiologic examination becomes very important. Direct radiography, knee-hip ultrasonography, echocardiography and laboratory and genetic studies are used for this purpose.

Treatment remains controversial. Affected patients are usually treated with anti-inflammatory drugs, methotrexate and biological agents. However, patients do not respond to medical therapy, although they may benefit from calcium and vitamin D supplementation for treatment of osteoporosis². Physiotherapy and surgical therapy may be beneficial for bone and joint deformities.

Surgical therapy may be inevitable for some patients who have pericarditis with or without pericardial effusion. There exist only a few case reports concerning surgery on these patients, so we are presenting here our surgical approach to pericarditis with pericardial effusion in a CACP syndrome patient.

Case Report

A seven-year-old boy complained of pain and erythema in his knees for the preceding two years. He was treated as juvenile rheumatoid arthritis with anti-inflammatory drugs. One year ago, he underwent a surgical operation, along with physiotherapy, for camptodactyly on his hands. Three months ago, echocardiography revealed a minimal pericardial effusion. We repeated the echocardiography and detected cardiac tamponade signs with pericardial effusion.

Laboratory findings were normal. Complete blood count, CRP and erythrocyte sedimentation rate values were normal. First, we performed a subxiphoid incision under local anesthesia. The pericardium was very thick and adherent to the heart, so we performed a full sternotomy under general anesthesia. We evacuated 100 ml of serous fluid. After harvesting the pericardium from the surface of the heart, we put in a

pericardial tube for drainage.

The postoperative period was uneventful. The mediastinal tube was removed on the second day, and the patient was discharged on the fourth day without any problems.

Pericardial biopsy revealed nonspecific signs of inflammation. Histopathological study of the fluid revealed nonspecific pericardial effusion.

After discharge, genetic studies were conducted and revealed a homozygous mutation in the *PRG4* gene, so the family was referred for genetic counseling. There was no parental consanguinity.

Discussion

Camptodactyly-arthropathy-coxa vara-pericarditis syndrome was first described in 1986³; the association between this syndrome and the proteoglycan 4 gene was detected in 1998⁴. Mutation of the gene responsible leads to synovial hyperplasia and loss of lubrication functions. CACP syndrome is a very rare disorder, so diagnosis may be challenging. Genetic studies are very important for differential diagnosis, given that the syndrome mimics rheumatologic disorders such as juvenile rheumatoid arthritis. Nonspecific joint deformities may be seen in childhood. Flexion deformity of the proximal interphalangeal joint of one or several fingers or abduction or internal rotation deformity of the hip joint are other components of joint involvement.

Anti-inflammatory therapy is not effective for these patients; however, most patients are receiving such therapy at the time of diagnosis. They occasionally have physiotherapy or surgical intervention in their medical history. Our patient had physiotherapy and surgery for the flexion deformity of his fingers (Fig. 1) one year ago; he also had coxa vara deformity (Fig. 2).

The prognosis of CACP syndrome is generally good, and patients do not have serious problems⁵. The disorder leads to noninflammatory pericarditis with pericardial effusion in 30% of affected patients, so echocardiographic examination becomes of considerable importance. Our patient had constrictive pericarditis and pericardial effusion, which was detected during periodic examination. He underwent operation due to cardiac tamponade.



Fig. 1. Direct radiography showing camptodactyly in the hands.



Fig. 2. Radiological view of coxa vara deformity.

In most CACP patients, the pericardial effusion is not large, so conservative therapy is sufficient. When clinical and radiological signs of tamponade are seen, intervention, including pericardiosynthesis or pericardiectomy, becomes inevitable. Although there have been only a few case reports concerning surgical intervention in CACP, when tamponade is detected, it should be resolved as soon as possible. Initially, pericardiosynthesis guided by echocardiography under local anesthesia may be effective for drainage of the fluid. Subxiphoid incision is a minimally invasive method without serious complications. When the pericardium is thick and adherent to the heart, median sternotomy may be the only choice, as was the case in our patient. We first tried the subxiphoid approach and then switched to median sternotomy to harvest the adherent pericardium from the surface of the heart. Pericardial fluid was serous

and amounted to 100 ml.

In the literature, the term “familial fibrosing serositis” has also been used for CACP syndrome. The syndrome may be seen in cousins, siblings and children born of consanguineous marriages⁶. In our patient, familial history was negative but there was a homozygous mutation in the disease gene. Genetic counseling is essential for family members.

Conclusion

Camptodactyly-arthritis-coxa vara-pericarditis syndrome is a very rare congenital disorder, such that differential diagnosis is difficult. It should be kept in mind in the case of children who have early childhood-onset joint deformities or arthritis. After diagnosis, echocardiographic examinations become important, in order to detect effusion and pericardial thickening. Surgery should be considered in these patients when signs of tamponade appear.

REFERENCES

1. Offiah AC, Woo P, Prieur AM, Hasson N, Hall CM. Camptodactyly-arthritis-coxa vara-pericarditis syndrome versus juvenile idiopathic arthritis. *AJR Am J Roentgenol* 2005; 185: 522-529.
2. Shaw NJ. Management of osteoporosis in children. *Eur J Endocrinol* 2008; 159: S33-S39.
3. Laxer RM, Cameron BJ, Chaisson D, Smith CR, Stein LD. The camptodactyly-arthritis-pericarditis syndrome. *Arthritis Rheum* 1986; 29: 439-444.
4. Bahabri SA, Suwairi WM, Laxer RM, Polinkovsky A, Dalaan AA, Warman ML. The camptodactyly-arthritis-coxa vara-pericarditis syndrome: clinical features and genetic mapping to human chromosome 1. *Arthritis Rheum* 1998; 41: 730-735.
5. Choi BR, Lim YH, Joo KB, et al. Camptodactyly, arthritis, coxa vara, pericarditis (CACP) syndrome: a case report. *J Korean Med Sci* 2004; 19: 907-910.
6. Bulutlar G, Yazici H, Ozdoğan H, Schreuder I. A familial syndrome of pericarditis, arthritis, camptodactyly, and coxa vara. *Arthritis Rheum* 1986; 29: 436-438.