

## Case Report

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## Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome: A rare childhood syndrome

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### Abstract

CACPS is the rare form of congenital autosomal recessive illness associated with mutation in proteoglycan 4 (PRG 4) gene. Here we are trying to present a case of 20 year old girl with complaints of pain in right hip, which is insidious in onset and progressive in nature which was radiating towards knee. The patient was having bow-leggedness since birth. Patient was operated and osteotomy was performed with angle plate fixation. Awareness regarding this syndrome should be increased in order to avoid confusion with other musculoskeletal disorders seen during childhood. Since Juvenile Idiopathic Arthritis is similar to CACPS it is clinically important to differentiate as both the conditions have different management protocols, in particular due to severe adverse effect profile associated with treatment of JIA.

**Keywords:** Camptodactyly-Arthropathy-Coxa-vara, Osteotomy, Bow-leggedness.

### INTRODUCTION

Camptodactyly-Arthropathy-Coxa-vara-Pericarditis Syndrome (CACP) is the rarest form of congenital autosomal recessive illness associated with mutation in proteoglycan 4 (PRG 4) gene characterized by early-onset camptodactyly and hyperplasia of the synovium with non inflammatory arthropathy, progressive coxa vara deformity and non inflammatory pericardial or pleural effusion affecting hips, wrist and small joints of hands. These characteristics mimics the most common rheumatic diseases hence are often misdiagnosed as juvenile idiopathic arthritis (JIA) <sup>1, 2</sup>.

For appropriate management careful assessment and early diagnosis of CACP syndrome is helpful. Laboratory investigations such as ESR, CRP and full blood count are usually normal in this syndrome while 30% of CACP patients are being reported with pericarditis. In both children and adults radiologic findings such as Ultrasonography and MRI are useful tools in assessing synovitis of metacarpophalangeal and wrist joints.<sup>3,4</sup>

Management remains controversial. Treatment is done with anti inflammatory drugs, methotrexate and biological agents. Sometimes, patients may not respond to medical therapy, but are benefitted with calcium and vitamin D supplementation for treatment of osteoporosis.<sup>5</sup> For treatment of joint and bone deformities physiotherapy and surgery are usually beneficial. Surgical therapy may be inevitable for some patients who have pericarditis with or without pericardial effusion.<sup>1</sup>

### CASE REPORT

A 20 Years old female patient presented with complaints of pain in right hip, which is insidious in onset and progressive in nature which was radiating towards knee. The patient was having Bow-leggedness since birth. Her present weight is 30kgs.

She denied cough, chest pain, fever, chills, rigors, and any neurological deficit involving upper extremities.

On physical examination her vital signs were fluctuating as shown in given table:

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**Table 1:** Physical examination

PARAMETERS	DAY 1	DAY 2	DAY 3	DAY 4	DAY 5	DAY 6	DAY 7
BP(mmHg)	120/70	100/70	90/80	130/80	90/70	110/80	120/70
Pulse bpm	120	96	92	98	87	88	94
Temperature 99.8 F	101.3 F	100 F	99F	99 F	98.7 F	99.7 F	99.7 F

Physical examination revealed

Tenderness (+)

ROM ( Range of Motion ): Flexion: upto 90°

Abduction : 15 °

Adduction : 15 °

On examination of spine showed following findings :

Lumbar lordosis(+)

Thorax Scoliosis (+)

Patient was having short stature and pigeon shaped chest as shown in figure



**Fig. 1:** X-RAY of chest spine and legs

**Past Medical History** :- patient as born with low birth weight and bow-leggedness, patient has slow growth development .

**Lab investigations:** Apart from the above mentioned physical examinations the patient underwent routine test as well as definitive tests.

**Routine Test:** complete blood picture revealed decrease Hb levels- 9.4 gm/dl as well as decrease RBC count -3.6 mill/cumm, a total WBC count of 7,000/cumm with a differential count of 17 % lymphocytes, and Neutrophil 78 %. Platelet count – 1.0 lakhs/cumm.- which revealed normocytic normochromic anaemia Electrolytes , ESR values were normal.

Patient was performed Chest X ray to rule out respiratory tract infections and was found to have increased bronchovascular margins noted in bilateral lungs field more prominent on right side as shown in figure.



**Fig. 2:** Chest PA view

Patient Thyroid Profile was found to be normal.

**Definitive Tests**

**x-ray** – X ray of hip revealed deformity that results in reduced angle between head and shaft of femur as shown in figure



**Fig. 3:** Pelvis AP View

X-ray of Skull revealed positional skull asymmetry, X-ray of fingers revealed comptodactyly given in figure



**Fig. 4:** Skull and hand APOBL

**Treatment:** Patient was given appropriate treatment as per physical examinations and lab investigations.

Patient was given cap Bacelac forte and Nebuliser –Budesonide and Duolin to treat respiratory tract infection and advised to perform osteotomy with angle plate fixation as shown in figure.



**Fig. 5:** Post Operative angle plate fixation

Post-operation the patient was given Inj .Amikacin 500mg/BD, Inj.metronidazole- 100ml/TID, Inj .Amoxicillin and clavulonic acid - 1.12gm/BD, Inj pantoprazole- 40mg/OD, T. Limcee- 1 tablet for 5 days and shifted to oral dosage form later

**Discharge medication:-** Patient was discharged after 10 days of appropriate treatment after ruling out the disease and given T.Limcee- 1 tab, cap Bacelac forte, T. Diclofenac -150mg for 1 week and advised for referral after 10 days.

Patient was also advised to perform physiotherapy and exercise regularly to improve movement and condition.

## DISCUSSION

CACP syndrome first described in 1986 is a genetic disorder which is autosomal recessive caused due to mutation of gene Proteoglycan-4" (PRG-4), which is a megakaryocyte stimulating factor gene encoding surface lubricant for joints and tendon.<sup>6</sup>

The pathophysiology of this illness is not well understood but Patients lack the glycoprotein 'lubricin' whose absence causes hyperplasia in the synovium, which is expressed not only in joints but also in pericardial and pleural cavities, liver, kidneys and skeletal muscles. Its formation involves transcription of the proteoglycan 4 gene (PRG4).<sup>7</sup>

CACP is a syndrome with Hallmark features of camptodactyly, arthropathy, coxa vara and pericarditis which may occur due to regulatory dysfunction in the proliferation of synovial and serosal cells. These all features are seen in our present case.

It may also include congenital cataracts. Pericarditis may be a presenting feature of this disease, fibrosis occurring in the pericardium and the tenosynovium may represent the end stages of processes similar to those occurring in the joint.<sup>8</sup>

Treatment includes extension relief from stress and strain but seems now to have been abandoned. Operative treatment is commonly done with the objective of rendering the hip Painless, Stable, functionally active and free from deformity or imminent shortening of the limb.<sup>9</sup>

## CONCLUSION

If radiographs of patients reveal an absence of erosion then CACP syndrome should be considered in all patients who present with non inflammatory arthropathy or JIA. Awareness regarding this syndrome should be increased in order to avoid confusion with other musculoskeletal disorders seen during childhood. Since Juvenile Idiopathic Arthritis is similar to CACPS it is clinically important to differentiate as both the conditions have different management protocols, in particular due to severe adverse effect profile associated with treatment of JIA.

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## Conflict of Interest

None Declared.

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