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CASE REPORTS

CAMPTODACTYLY ARTHROPATHY COXA VARA PERICARDITIS SYNDROME: A MIMICKER OF JUVENILE IDIOPATHIC ARTHRITIS- ZEBRA AMONGST HORSES

Avinash Sharma[1], Yamini Sharma[2].

[1]Pediatric clinical immunology and Rheumatologist, Dr Rajendra Prasad Government Medical College, Tanda, Kangra, Himachal Pradesh, India,

[2]Department of Pediatrics, Dr Rajendra Prasad Government Medical College, Tanda, Kangra, Himachal Pradesh, India.

ABSTRACT

Juvenile idiopathic arthritis (JIA) is a common rheumatological condition of childhood that clinically presents as pain, swelling, morning stiffness and restricted function of the affected joints. The International League of Associations for Rheumatology (ILAR) criteria are used for classification of JIA.[1] There is no laboratory tool for diagnosis, which is essentially clinical. Certain noninflammatory disorders may present with joint swelling, contractures or deformity with minimal or no pain and no signs of inflammation. These non-inflammatory arthropathies mimic JIA.[2] Thus, diagnosis often gets delayed, resulting in delay in appropriate management. The reasons for misdiagnosis appear to be rarity of these conditions and lack of awareness. We report a case of Camptodactyly-Arthropathy-Coxa Vara-Pericarditis (CACP) Syndrome who was diagnosed at the age of 13 years.

ARTICLE HISTORY

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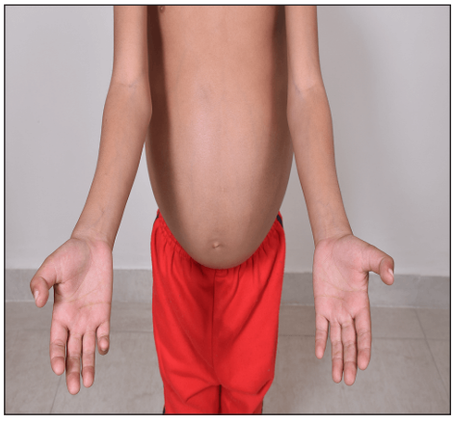
KEYWORDS

Non-inflammatory, arthritis, arthropathy.

Case Report

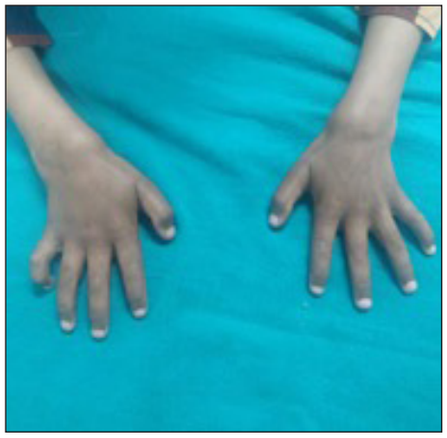
A 13 years, boy presented to Pediatric Rheumatology Clinic with complaints of pain, swelling and restriction of movements of bilateral knee joints since the age of 3 years and abdominal distension from last 6 months. Patient was second child in the family and was born of non-consanguineous marriage. He had had multiple hospitalizations in past and was prescribed oral steroids on multiple occasions for inflammatory arthritis. On examination, proportionate short stature with camptodactyly in hands and flexion deformity of bilateral elbow joints was noted (Figure 1, 2)

Figure 1. Showing gross ascites and cubitus valgus.



Address for Correspondance: Dr. Avinash Sharma, Government Medical College, Tanda, Kangra, Himachal Pradesh, India.
Email: avieku9307@gmail.com

Figure 2. Showing camptodactyly.



Also detected was free fluid in abdomen, bilateral dull note on chest percussion and muffled heart sounds on auscultation. Erythrocyte Sedimentation Rate (ESR) and C Reactive Protein (CRP) were not elevated. 2-dimensional echocardiography revealed constrictive pericarditis. X-ray of hip joints showed sclerosis and irregularity of acetabular margins with femoral neck shortening. Clinical possibility of CACP was considered in view of non-inflammatory arthropathy, classical physical and radiological findings.

Clinical exome sequencing showed homozygous 7 base-pair deletion in exon 7 of PRG4 gene resulting in frameshift and premature truncation of protein at codon 1085 (p.Ser1085Ter;ENST00000445192.2) and thus diagnosis was confirmed and child was initiated on spironolactone. Child underwent pericardiectomy and now is doing well after 36 months of follow-up.

Discussion

CACP (Camptodactyly-Arthropathy-Coxa vara-Pericarditis syndrome) is a non-inflammatory arthropathy that mimicks JIA.[3] It is an uncommon, autosomal recessive condition resulting from mutation in PRG4 gene. This gene encodes for proteoglycan 4, the predominant lubricant in joints. Camptodatyly is a universal finding in CACP along with Arthropathy of large joints with periarticular osteopenia without erosions. There is coxa vara, short femur neck and flat femoral head with pericarditis seen in about 30% of cases.[4] There is no specific treatment for this condition.[3]

Progressive Pseudo-rheumatoid dysplasia, arthritis in mucopolysaccharidosis and idiopathic multicenteric osteolysis are few more examples of non-inflammatory arthropathies which need to be differentiated from inflammatory arthritis.

Progressive Pseudo-rheumatoid dysplasia closely mimics JIA. It is caused due to a mutation in WISP3 gene that is transmitted in an autosomal recessive pattern. Like JIA, child presents with progressively deforming arthropathy, largely involving small peripheral joints resulting in contractures.[5] Characteristically, pain and other signs of inflammation are absent and laboratory parameters of inflammation are not elevated. Radiologically, dysplastic changes are noted in the involved joints.

Mucopolysaccharidoses (MPS) are inborn disorders of the metabolism of glycosaminoglycan.[3] Musculoskeletal manifestations in form of joint contractures are prominent in all forms of MPS which are also seen in inflammatory arthritis and thus may result in misdiagnosis. Other clinical features like coarsening of facies and certain radiographic findings, such as characteristic dysplasia and dysostosis multiplex, should make one suspect MPS.[6]

Idiopathic osteolysis is a rare inherited heterogeneous group of disorders.[3] Young children present with pain, bony deformities and restricted movements of joints which simulates arthritis. X-ray and computed tomography (CT) scan demonstrate lytic lesions.

Conclusion

Increased awareness of non-inflammatory arthropathies helps in differentiating them from JIA and helps in timely diagnosis. In our index case, it took ten years for diagnosis to be established. Genetic disorders with musculoskeletal involvement that mimic chronic polyarthritis should be considered in differential diagnostics of JIA. These disorders closely mimic JIA. Detailed history, examination and imaging will render a timely diagnosis and avoid pitfall of inappropriate therapy. Absence of pain, morning stiffness, normal ESR and CRP, characteristics radiological features and some specific clues like camptodactyly in CACP syndrome are often helpful in distinguishing these disorders from JIA.

One must always look for these clues that help identify zebra from amongst horses.

Compliance with Ethical Standards

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Conflict of Interest None

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