Acta Scientific Orthopaedics (ISSN: 2581-8635)

Volume 5 Issue 12 December 2022

Case Report

Treatment of a Case of Intra-arterial Calcification of Popliteal Artery in a Young Boy

Gopala Krishna B. A*

Department of Orthopaedics, Manipal Hospitals, Bangalore, Karnataka, India

***Corresponding Author:** B. A. Gopala Krishna, Department of Orthopaedics, Manipal Hospitals, Bangalore, Karnataka, India.

DOI: 10.31080/ASOR.2022.05.0630

Received: September 06, 2022

Published: November 11, 2022

Abstract

Introduction: ACDC is a rare autosomal recessive genetic disorder caused by mutation in the NT5E gene, which results in extensive calcification of vessels below the diaphragm, notably the large lower extremity arteries and small joints of hands and feet [3,4]. Also known as "calcification of joints and arteries" (CALJA) syndrome, the late-onset profound calcification spares arteries in the trunk and the coronary circulation.

Case Report: We report the case of, an 11-year-old boy, who presented with abrupt and abnormal calcific deposits at various anatomical location with pain and limitation of the movements of the involved region. The symptoms were not controlled with standard anti-inflammatory medications with other supportive measures. The symptoms were significant enough to limit his activities of daily living.

Conclusion: ACDC is a rare genetic disorder with limited knowledge to many practicing physicians and surgeons. The symptoms are debilitating depending on the anatomical region affected. Inadvertent usage of medication or intervention can lead to complications or undesired outcomes. Hence a detailed study of such cases with standard treatment recommendations are necessary. This case report shows a favourable outcome with the treatment approach that was instituted. However study of a reasonable number of similar cases can throw better light in understanding and managing such rare disorder.

Keywords: Intra-arterial Calcification; NT5E Gene Mutation; ACDC; CALJA Syndrome; CD 73 Deficiency

Abbreviations

ACDC: Arterial Calcification Due to Deficiency of CD73; CALJA: Calcification of Joints and Arteries; DP: Dorsalis Pedis; TP: Tibialis Posterior; ATP: Adenosine Triphosphate; AMP: Adenosine Monophosphate; TNAP: Tissue Nonspecific Alkaline Phosphatase

Case Presentation

11 year old boy presented with persistent and significant heel pain that was not controlled with standard medications. He was the 1st child of his parents and was born out of nonconsanguineous marriage and was born by normal vaginal delivery at term. Vaccinated appropriately for his age.

The treating surgeon at his place found a calcific deposit in the heel pad on routine x ray. CT scan showed similar calcific deposit in the great toe distal phalanx and was suggestive of possible systemic calcium deposit disorder. A biopsy from the lesion was inconclusive. The treating surgeon did excision of the calcific deposit. Soon the boy developed similar calcific deposit in the other heel pad.

The boy was referred to us when he developed sudden onset left knee pain. He had significant knee pain with flexion deformity. However range of motion of the knee was fair and painless. This raised suspicion of some extra articular pathology. Routine X ray of the knee showed serpentine calcification of popliteal artery. The DP pulse was absent on left side and feeble on right side and TP

pulse was feeble on both sides. Since he also had claudication pain in both lower limbs detailed evaluation with CT angiogram of both lower limbs was done. This revealed extensive calcification from common iliac artery downwards on both sides.

Laboratory investigations revealed normal blood counts with erythrocyte sedimentation rate of 20 mm/1st h. Rheumatoid factor and anti-cyclic citrullinated peptide were negative. Serum alkaline phosphatase was within normal limits. Serum calcium level was 9.1 mg/dL and serum phosphorus level was 4 mg/dL.

The radiological picture along with arterial calcification and clinical presentation seemed to be highly suggestive of arterial calcification due to deficiency of CD73 (ACDC) [1,4]. Due to financial constraints, detailed genetic evaluation could not be carried out. However there was no family history of claudication pain or similar complaints in any of his family members.

The boy was started on Ibandronic acid 75 mg once a month for 6 months, Cilostazol (to improve symptoms of claudication) daily for 2 months along with Ecospirin 75 mg daily for 3 months. The boy was asked to be as active as possible to encourage formation of collaterals to prevent ischaemia.

On follow up evaluation after 12 weeks, the knee pain subsided completely, there was no claudication symptoms and no flexion deformity of knee. Repeat X ray of the involved knee showed significant dissolution of the popliteal artery calcification and Left heel pad calcification. He was evaluated every 8 weeks for any formation of new calcific deposit and resolution of old deposit. At the end of 1 year from the onset of initial symptoms, there was no recurrence of either symptoms or new calcific deposit and there was significant dissolution of initial calcific deposit in left popliteal artery and left heel pad.

Discussion

NT5E gene encodes for CD73, a membrane-bound ecto 5'-nucleotidase, responsible for extracellular Adenosine Triphosphate (ATP) metabolism throughout the body. Adenosine monophosphate (AMP) and pyrophosphate are formed from ATP by the action of ectonucleotide pyrophosphatase-phosphodiesterase. Normally, CD73 binds to AMP and converts it to adenosine and inorganic phosphate. Tissue nonspecific alkaline phosphatase (TNAP) is responsible for the breakdown of pyrophosphate throughout the body. Calcification process depends on the levels of pyrophosphate, which is crucial for the inhibition of calcification [2]. TNAP is inhibited by adenosine. NT5E mutation results in little or no functional CD73, ultimately resulting in excessive calcification. The predilection for lower extremity arteries is attributed to the pattern of distribution of adenosine receptors. Intermittent arthritis resembling basic calcium phosphate crystal deposition disease and early-onset osteoarthritis are found to be characteristics of ACDC. [2]. Bisphosphonates maybe beneficial in patients with CD73 deficiency, as they are pyrophosphate analogs and hence can inhibit tissue calcification.

Figure 1: Heel pad calcification before the start of treatment.

Figure 2: Post treatment, heel pad calcification has completely n resolved.

Figure 3

Conclusion

ACDC is very less known entity. There is no established treatment protocol for the same. However our limited experience with this single case had a favourable outcome with the treatment strategy followed. But the safety of usage of Ibandronic acid in younger kids is not very well established also the recommended dose and the duration of treatment needs to be understood. Risk factors for recurrence and its prevention has to be evaluated thoroughly.

Conflict of Interest

No financial aid from any source was sought during the process of this case report study.

Bibliography

1. Cudrici CD., *et al*. "Multifocal calcific periarthritis with distinctive clinical and radiological features in patients with CD73 deficiency". *Rheumatology* (Oxford) 61.1 (2021): 163-173.

2. St Hilaire C., *et al*. "NT5E mutations and arterial calcifications". *The New England Journal of Medicine* 364.5 (2011): 432-442.

3. Naomi Ichikawa., *et al*. "Arterial Calcification Due to Deficiency of CD73 (ACDC) As One of Rheumatic Diseases Associated with Periarticular Calcification". *Journal of Clinical Rheumatology* 21.4 (2015): 216-220.

4. Luis Balmore Gutierrez., *et al*. "Arterial calcification due to CD73 deficiency (ACDC): imaging manifestations of ectopic mineralization". *Skeletal Radiology* 45 (2016): 1583-1587.