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Case Report

Imaging insights into ACVR-1-linked ossification syndrome

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ABSTRACT

Fibrodysplasia ossificans progressiva is a rare autosomal dominant genetic disorder (ACVR-1 gene) with a prevalence of 1 in 2 million births. It is characterized by heterotopic progressive ossification in connective tissues and between osseous structures. Congenital malformation of the great toes is a pathognomic association. Herein this repost presents a case of a thirteen-year-old girl who presented with difficulty in walking for one year. She had short bilateral great toes and had an abnormal gait. Radiographs followed by computed tomography (CT) of the bilateral hip joint showed soft tissue ossification around the hip joint with bilateral coxa valga. Magnetic resonance imaging of the bilateral hip was also done, which showed intramuscular edema in the left gluteus medius and maximus. Early diagnosis and identification of this disease with imaging is important to prevent unnecessary biopsies and surgical intervention.

Keywords: Heterotopic ossification, Genetic disorder, Hallux valgus, Progressive deformity, Computed tomography

INTRODUCTION

Fibrodysplasia ossificans progressiva is a rare autosomal dominant genetic disorder (ACVR-1 gene) with a prevalence of 1 in 2 million births.[1] It is characterized by heterotopic progressive ossification in connective tissues and between osseous structures. Congenital malformation of the great toes is a pathognomic association.[2]

We present a case report highlighting the imaging aspects of this rare disease.

CASE REPORT

Clinical history

A thirteen-year-old girl presented with bilateral hip joint pain for the last one and a half years. The pain was insidious in onset and gradually worsened. She also experienced difficulty sitting and walking for the past year. There was no history of trauma or prior surgery.

Imaging findings

Conventional radiographs of both hips showed ossification bands around the joints, extending from the greater trochanter to the ischium. A mild reduction in joint space was also observed (Figure 1).

Figure 1: Radiograph of the bilateral hip shows ossification bands around the hip joints extending superomedially from the greater trochanter. There is a mild reduction in the bilateral hip joint space.

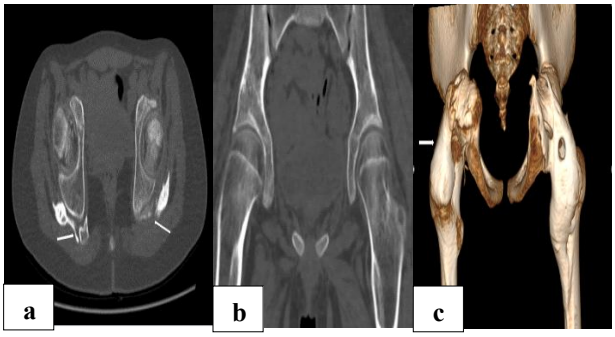

Figure 2: Computed tomography image of bilateral hips: (a) axial image shows heterotopic ossification in the posterior intramuscular compartment around the bilateral ischium and the ossification bands show displaced fracture (pseudo-exostosis) within (white arrows), (b) coronal image shows a mild reduction in the bilateral hip joint space and the bilateral femoral neck appears shortened and broad in appearance with a widened neck-shaft angle. (coxa valga Rt – 149.4 degrees, Lt –160 degrees), and (c) surface shaded three-dimensional image of the bilateral hip demonstrates an ossification band (white arrow) between the bilateral greater trochanter and ischium.

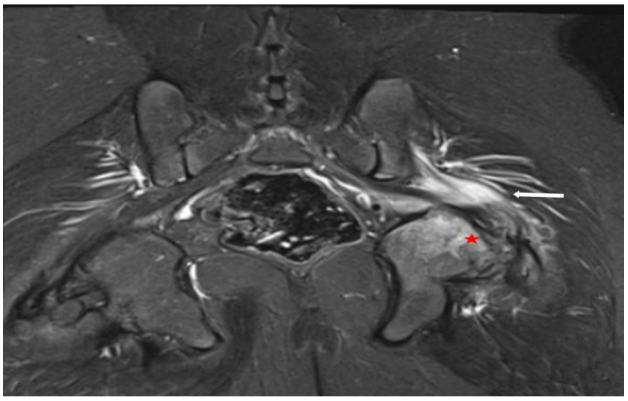

Figure 3: Magnetic resonance imaging of the bilateral hip (coronal TIRM sequence) showing intramuscular edema in left gluteus medius and maximus (white arrow) with bone marrow edema in left ischial tuberosity (red arrow).

Computed tomography (CT) scans of both hips revealed heterotopic ossification bands in the posterior intramuscular compartment of the thigh, extending from the greater trochanter posteriorly, superiorly, and medially to the ischium. The femoral necks were shortened and broad, with an increased neck-shaft angle, indicative of coxa valga (Figure 2).

Magnetic resonance imaging (MRI) of the hips revealed intramuscular edema in the left gluteus maximus, medius, and ischial tuberosity, along with heterotopic ossification in the posterior intramuscular compartment (Figure 3). CT scans of bilateral foot showed hallux valgus with shortened proximal phalanges of great toes. (Figure 4)

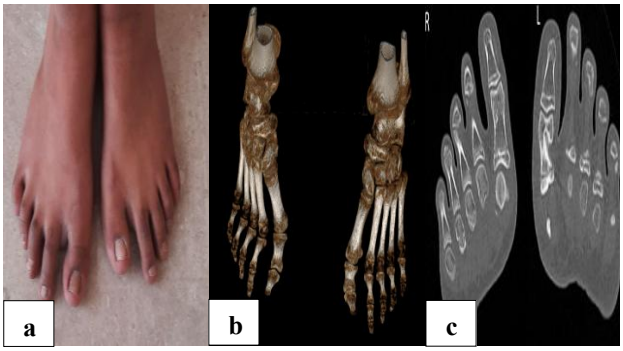

Figure 4: Bilateral hallux valgus and short great toes: (a) clinical view, (b) CT images of the bilateral foot show shortening of the proximal phalanx of the great toes seen (left > right), with bilateral hallux valgus, and (c) 3D reconstructed image of the bilateral foot.

DISCUSSION

Background

Fibrodysplasia ossificans progressiva (FOP) is a rare, autosomal dominant genetic disorder characterized by progressive heterotopic ossification in connective tissues and skeletal muscles.

Genetics

A mutation in the Activin receptor type IA (ACVR1) gene, which encodes a bone morphogenic protein (BMP) type I receptor, causes FOP.[3]

Pathogenesis

Mononuclear cell invasion in skeletal muscle leads to severe edema, replacing muscle tissue with fibroproliferative tissue that eventually matures into heterotopic bone via enchondral ossification.[4]

Clinical perspective

The age of onset varies from 6 months to 13 years, with a mean of around 5 years.[5] The disease features episodic joint swelling (flare-ups) that promote ossification in muscles, tendons, ligaments, and aponeuroses, leading to joint ankylosis and deformity.

Complications include early degenerative joint disease, temporomandibular joint ankylosis, ossification of ear ossicles, and scoliosis. Patients often become severely disabled and socially impaired. Death typically occurs by age of forty years due to cardiopulmonary failure.[6]

Hallux valgus with short toes is a hallmark congenital deformity of FOP.[7] Other abnormalities include short thumbs, clinodactyly, and vertebral anomalies.

Imaging perspective

Conventional radiographs can identify heterotopic ossification but are limited in detecting early inflammatory stages and in assessing the full extent of disease.

Ultrasound is useful during the inflammatory phase for detecting soft tissue edema.

CT provides a detailed three-dimensional visualization of disease extent, joint fusion, and progression.

MRI helps identify pre-ossification lesions, which appear as low signal on T1-weighted sequences and high signal on T2-weighted sequences, with muscle edema showing hyperintensity on T2-weighted sequences.[8]

Bone scintigraphy is highly sensitive for the early detection of heterotopic ossification.

Positron emission computed tomography (PET-CT) is utilized during flare-ups, with increased FDG uptake indicating heightened metabolic activity.

Confirmation is achieved through ACVR1 gene analysis.

Treatment and outcome

A multidisciplinary approach, including patient education, pain management, and injury prevention, forms the core of management. Medications such as NSAIDs, corticosteroids, and high-dose bisphosphonates are given in acute flare-ups. Surgical removal of heterotopic bone is generally avoided, as it can worsen the disease.[9,10]

CONCLUSION

Early diagnosis and identification of fibrodysplasia progressiva ossificans with imaging is important to prevent unnecessary biopsies and surgical intervention. Biopsy is contraindicated and will cause a disease flare-up. Congenital malformation of the great toe is an associated pathognomic feature and thus can aid in diagnosis.

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