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Case Report

A rare case of progressive pseudorheumatoid dysplasia

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ABSTRACT

Progressive pseudorheumatoid dysplasia is a rare, autosomal recessive, noninflammatory musculoskeletal disorder caused by mutations occurring in the WNT1- protein present in 3 gene. Usual presentation is arthralgia, joint stiffness, contractures with primary involvement in joint cartilage. We present a 13-year-old female child presented with history of short stature, bowing of legs and curving of spine and inability to walk normally since the past two years. The symptoms have been progressively worsening for the past one year. Progressive pseudo rheumatoid dysplasia is a rare form of spondyloepimetaphyseal dysplasia and is clinically misdiagnosed as juvenile idiopathic arthritis. Any patient presenting with musculoskeletal symptoms should be evaluated appropriately to start appropriate therapy in time.

Keywords: Pseudorheumatoid dysplasia, Short stature, WISP3 gene

INTRODUCTION

Progressive pseudorheumatoid dysplasia (PPD) is defined by the predominance of articular cartilage involvement and progressive stiffening and enlargement of the joints without inflammation. Permanently bent fingers, larger finger and knee joints, and a narrowing of the space between the bones at the hip and knee joints are other indications and symptoms that emerge over time. A prevalent issue among adolescents is hip pain. Affected people have flattened, improperly formed (beaked) spine bones called platyspondyly, which results in an abnormal front-to-back curvature of the spine called kyphosis and a short torso.[1] The WISP3 gene (WNT1-inducible signalling pathway protein 3) encoded on chromosome 6q21 is the cause of this illness. By controlling the chondrocytes' expression of type II collagen and aggrecan, WISP3 is crucial for preserving cartilage integrity. As a result, it is essential for preserving cartilage and forming bones.[2-4] Gait and posture are affected as the condition progresses, and there is severe morbidity. It causes significant joint contractures, gait abnormality, scoliosis, or kyphosis, which results in abnormal posture and significant morbidity, beginning between the ages of 3 and 6 years with involvement of the interphalangeal joints as the initial clinical presentation and, over time, involving the large joints and the spine.[5]

CASE REPORT

A 13-year-old female child presented with history of short stature, bowing of legs and curving of spine and inability to walk normally since the past two years. The symptoms have been progressively worsening for the past one year. There was no history of trauma and pain. She was born by normal vaginal delivery to consanguineous marriage. Her Younger sister is 9 years old and of normal height. Second degree male relative- suffers from similar problem. Her developmental milestones-were normal. Attained menarche 2 years ago, menstrual cycle- normal with regular flow. Her vitals were normal.

Head to toe examination

Medium built but short stature, normal intelligence, no dysmorphic facies, upper trunk- short, knee- genu varum and spine- scoliosis, distal interphalangeal joint- widened.

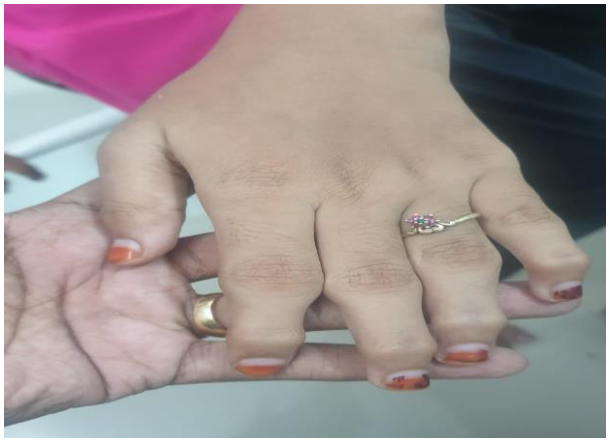

Figure 1: Swelling of the proximal and distal interphalangeal joints of the left hand.

Anthropometry

Height: 129 cm (<3rd centile), weight: 27.8 kg, expected height for age was calculated to be 148 cms, arm span: 140 cms, upper segment to lower segment ratio was 0.8:1, mothers height was 156 cms fathers' height was 163 cms, mid parental height was 156 cms.

Investigations

Laboratory findings

Normal erythrocyte sedimentation rate (ESR), C-reactive protein (CRP), and complete blood count (CBC) were reported.

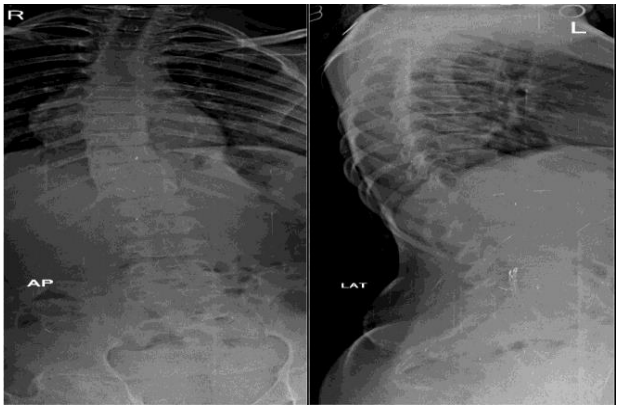

Figure 2: X-ray spine.

Radiologic features

Bone age assessment- X ray bands with wrist – epiphyses appears wider than the metaphyses, physeal fusion evident in the distal phallanges and metacarpals. The proximal phallanges also show minimal degree of physeal fusion. The bone age falls b/w 13 to 15 years.

In the spine, mild degree of platyspondyly seen in thoracic and lumbar vertebrae (especially in L3, L2, L1, T12, T11, T10), mild genu valgum bilaterally (right>left).

In the pelvis, flattened right femoral epiphyses was reported.

In the knees, secondary osteoarthritic change was reported.

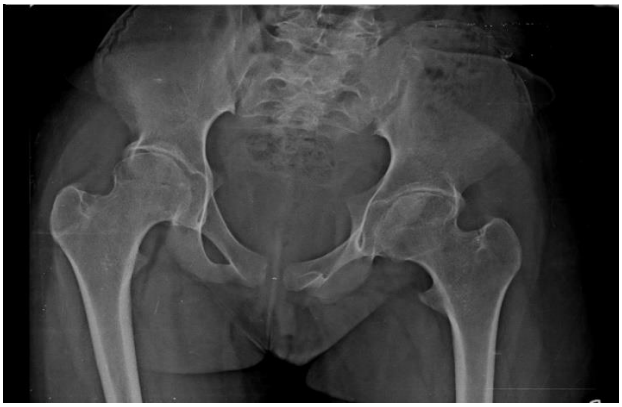

Figure 3: X-ray of hip.

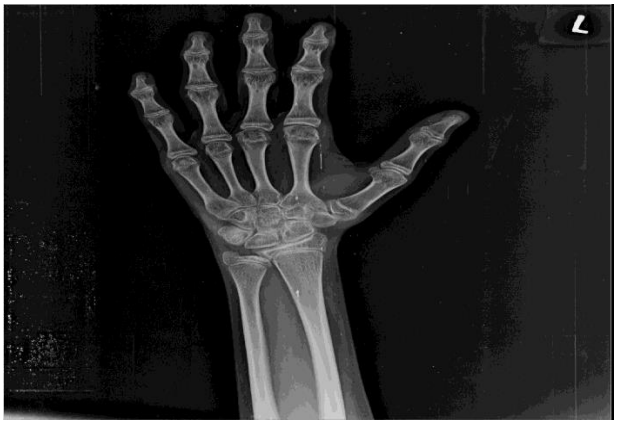

Figure 4: X-ray of wrist.

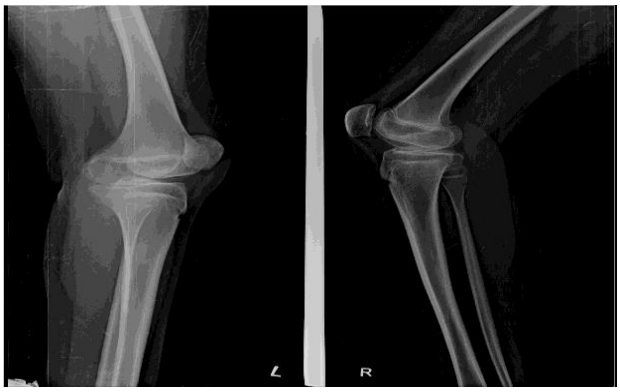

Figure 5: X-ray knee joint.

Management

Supportive treatment, NSAIDs for pain due to secondary osteoarthritis, Physiotherapy, activity modification and walking aids for large joint stiffness. An occupational therapist treats small joint arthopathy and may suggest assistive technology. Bracing can be used to treat minor cases of kyphosis and scoliosis. Prevent immobility, such as casting. Since these patients do not respond to antirheumatic medications, there will be no clinical advantage. Although there is no known treatment for PPRD, it may be managed with painkillers and early joint replacement surgery for the hip and knee.[5] Spondyloepiphyseal dysplasia with platyspondyly as an early finding and a lack of damaging joint erosions are radiological hallmarks of JIA that set it apart from PPRD.[4,5]

DISCUSSION

PPD, a distinctive form of spondyloepiphyseal dysplasia (SED) characterised by predominant involvement of articular cartilages.[6] It is an autosomal recessive disorder. It is caused by mutations in the WISP3 gene, which encodes for a protein expressed in synoviocytes and chondrocytes that plays a major role in bone and cartilage development.[7] The disease is typically silent at birth and early childhood. It usually manifests between the ages of 3 and 8 years, but the diagnosis is most often made in the second decade.[8] PPD, though a rare disease, should be kept in mind in the differential diagnosis of juvenile idiopathic arthritis, to prevent delayed diagnosis and to begin early rehabilitation thus avoiding potentially serious late morbidity.[9] Although radiologic examination has high accuracy in diagnosing PPRD, the definitive diagnosis is established by molecular genetic testing which identifies *WISP3* gene on.[10] Genetic testing and counselling should be offered to these families to plan for the next generation.

CONCLUSION

In conclusion, patients presenting with musculoskeletal symptoms should be evaluated appropriately to prevent inappropriate therapy, and to start appropriate therapy in time.

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