Case Report

Mucopolysaccharidosis type I-Hurler's syndrome

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Abstract

Mucopolysaccharidosis I (MPS I) is a rare inherited disorder that belongs to a group of clinically progressive disorders and is caused by the deficiency of the lysosomal enzyme, α1 -iduronidase. MPS I has been recently classified into a severe (Hurler syndrome) and an attenuated type (Hurler-Scheie and Scheie syndromes).
Keyword: α1 –iduronidase, Hurler-Scheie syndrome.

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INTRODUCTION

Hurler's syndrome is rare autosomal recessive disorder of mucopolysaccharide metabolism that leads to excessive lipoid accumulation in central nervous system and other viscera. Its incidence is 1:100000 of births. The excessive muccopolysaccharides excreted in urine are dermatan sulfate and heparan sulfate. Their synonym includes lipochondrodystrophy, Gargolism, osteochondrodystrophy, dysostosis multipex.

CASE HISTORY

3 years old male child presented with short stature (stunted growth), coarse facial features, limping and spinal deformity. Patient was subjected for radiograph of thoracic-lumbar spine AP and Lateral view, Both Hand AP view, skull AP and lateralview. Radiograph revealed

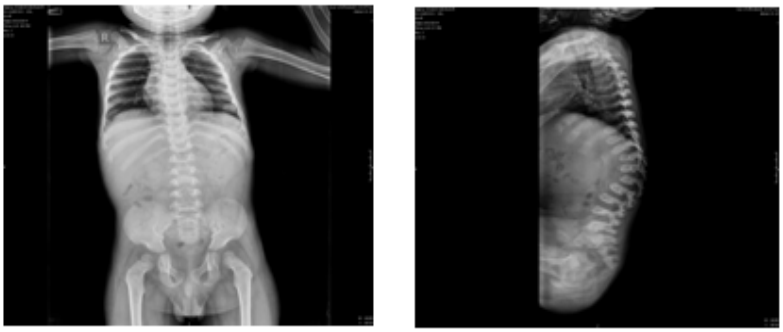

Figure 1: Thoraco-lumbar AP and Lateral view-> kyphosis of spine, Lower thoracic (T9 toT12) and upper lumbar vertebrae shows anterio-inferior beaking, appears small in antero-superior aspect, rest visualized thoracic vertebrae appears oval in shape. Visualized lower ribs appear wide anteriorly producing spatulated appearances. illi are flared, with obliquely directed acetabular roof

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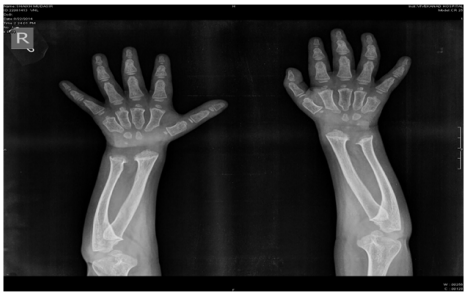

Figure 2

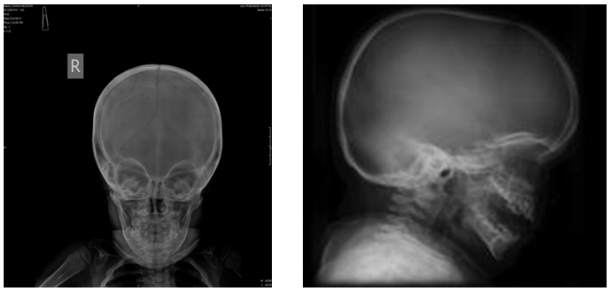

Figure 3

Legend

Figure 2: Both Hands AP view-> Metacarpals and phalynges are short, wide producing triradent hand.Bullet shaped metacarpals

Figure 3: Skull APand lateral view-> Macrocephaly, frontal bossing, calvarial thickening

Findings suggestive of Mucopolysaccharidosis 1-Hurler's syndrome.

DISCUSSION

Mucopolysaccharidosis-I (MPS I) is a lysosomal storage disorder inherited as an autosomal-recessive condition and is caused by a deficiency of the lysosomal enzyme α1 -iduronidase. This results in the progressive accumulation of glycosaminoglycans (GAG) within the lysosomes, leading to multiorgan dysfunction and damage[1]. Patients affected with MPS I are unable to degrade the GAG, dermatan sulfate, and heparan sulfate, which provide structural support to the extracellular matrix and cartilaginous structure such as joints and heart valves[2]. Patients are usually normal at birth and remain so until after the first year of life .Facial features then begin to coarsen, with the development of large head, wide set eyes (hypertelorism), sunken nose, large lips and protruding tounge. Corneal opacities develop and teeth are short and malformed. Mental retardation, deafness gradually develops. Hepatosplenomegaly, protruberant abdomen, umbilical and inguinal hernias are common. Eventually the patients become dwarfed. A severe dorso-lumbar kyphosis develop.[3] MPS I has been classified into two broader groups, severe MPS I (Hurler Syndrome) and attenuated MPS I (Hurler-Scheie and Scheie syndromes).[5] The greatest variability is observed in individuals with the attenuated MPS I. Onset is usually between ages three and ten years. Although pyschomotor development may be normal in early childhood, individuals with attenuated MPS I may have learning disabilities. The rate of disease progression and severity can range from serious life threatening complications (leading to death in the second to third decades) to a normal life span with significant disability and discomfort from progressive severe restriction in the range of motion of all joints. Hearing loss and cardiac valvular disease are common. The diagnosis of MPS I relies on the demonstration of deficient activity of the lysosomal enzyme α-L-iduronidase in peripheral blood leukocytes, cultured fibroblasts, or plasma. Glycosaminoglycan (GAG) (heparan and dermatan sulphate) urinary excretion is a useful preliminary test.[6] Other radiological findings includes –premature closure of sagital and lambdoid sutures. Hydrocephalus is common. Sella turcica is enlarged and J shaped. Dens hypoplasia resulting into atlantoaxial subluxation. In Pelvis illi are flared with obliquely directed acetabular roof. Coxa vera or vulga is common. Varus deformity of humerus is characteristic.[7] On sonography hepatomegaly may be seen.

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