



Case Series

Hurler Syndrome: A report of three cases

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Abstract

The mucopolysaccharidoses (MPS) are a group of inherited metabolic disorders that result in widespread skeletal, visceral, and mental abnormalities. A defect in metabolic degradation leads to the storage of mucopolysaccharide macromolecules in the nervous system and other body tissues. The MPS are classified into various types, and there are additional diseases, such as the mucolipidoses and a gangliosidosis, that demonstrate similar clinical and radiologic findings. We hereby present 3 cases of Hurler Syndrome, Type I mucopolysaccharidoses, and enumerate their radiological characteristic radiological findings.

Key words

Mucopolysaccharidoses, Hurler Syndrome, Skeletal changes.

Introduction

Hurler syndrome is one of the mucopolysaccharidosis (MPS type IH). It carries an autosomal recessive inheritance and manifests in the first years of life. It is clinically characterized by mental retardation, corneal clouding, deafness and cardiac disease, with death resulting in first decade of life, often from cardiac disease. The diagnosis is made by combining the clinical findings with the characteristic radiological findings. We hereby present 3 cases of Hurler Syndrome.

Case series

Three siblings of age 12 year, 8 year and 7 year were referred to Department of Radiology, Dhiraj General Hospital with complaints of intellectual and learning disability, short stature and growth retardation, thick, coarse facial features with low nasal bridge, Joint stiffness and loss of physical skills. Multiple X-rays were taken and showed macrocephaly, J-shaped sella, atlanto-axial subluxation, shortening and widening of long bones, pointing of proximal metacarpals, widening of anterior ribs (oar shaped/ paddle ribs), hypoplastic vertebra at thoracolumbar junction, anterior inferior



vertebral body beaking, thoracolumbar kyphosis which are characteristic findings of Hurler Syndrome. (Photo – 1 to 6)

Photo – 3: X-ray skull Lateral view: J - Shaped sella and macrocephaly.

Photo – 1: Clinical photograph of patients.



Photo – 2: X-ray skull AP view: Macrocephaly.



Photo – 4: X-ray both hand AP view: Proximal pointing of metacarpals, mildly angulated radius and ulna.



Discussion

Hurler's syndrome is a rare autosomal recessive disorder of mucopolysaccharide metabolism that leads to excessive lipid accumulation in the central nervous system and other viscera. It occurs in approximately 1/100,000 births. The excessive mucopolysaccharides excreted in the urine are dermatan sulfate and heparin sulphate [1].

Patients are usually normal at birth and remain so until after the 1st year of life. Facial features then begin to coarsen, with the development of a large head, widely set eyes (hypertelorism), a sunken nose, large lips, and a protruding tongue. Teeth are short and malformed. Mental deterioration ensues and deafness gradually

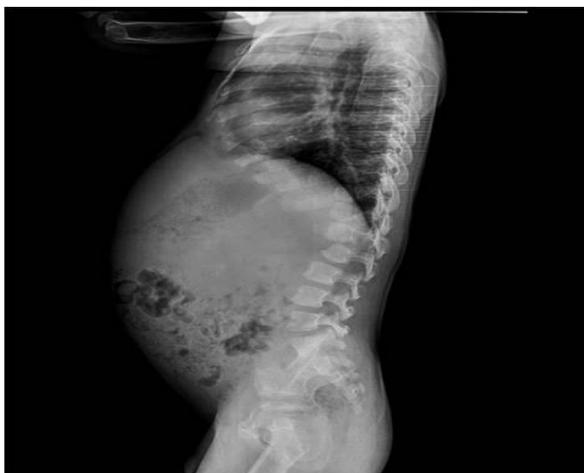


develops. Hepatosplenomegaly, umbilical and inguinal hernias are common. As physical development ceases in the early years of life, affected individuals become dwarfed. A severe dorsolumbar kyphosis develops, along with multiple flexion contractures. The hands are trident and sometimes clawed. Death usually occurs in the 2nd decade, often following pneumonia or cardiac failure. Enzyme replacement therapy has been developed as a potential therapy for some patients with MPS-I [2, 5].

Photo – 5: X-ray foot AP view: Shortening and widening of bones, stubby phalanges.



Photo – 6: X-ray D-L spine lateral view: L 2 small in size and posteriorly placed with inferior beaking.



Radiological changes seen in the skull include macrocephaly, frontal bossing, calvarial thickening, and premature closure of the sagittal and lambdoid sutures. Hydrocephalus is common. The sella turcica is enlarged and J shaped. Often, the facial bones are small and the mandibular angle is widened. Spinal changes are fairly typical. A thoracolumbar kyphosis develops secondary to vertebral body hypoplasia. The lower thoracic and upper lumbar bodies are small in their anterosuperior aspect and may be beaked inferiorly. The remaining vertebrae may be oval owing to convexity of the upper and lower surfaces [3]. The pedicles are often long and slender. Atlantoaxial subluxation has been reported as well as upper cervical instability. The ribs are overly wide, with tapered ends, producing a paddle or spatulated appearance. The ilia are flared, with obliquely directed acetabular roofs. Coxa valga or vara is common. Varus deformity of the humerus is characteristic. The tubular bones have widened diaphyses; this is more obvious in the upper extremities than in the lower. Often, the metacarpals and phalanges are short and wide, producing a trident hand. Osteoporosis is a frequent finding [4].

Conclusion

We concluded that Hurler syndrome is a rare inherited disorder of metabolism and is one of the lysosomal storage diseases referred to as mucopolysaccharidosis (MPS) that produce skeletal abnormalities, collectively termed “dysostosis multiplex”. It produces characteristic clinical and radiological findings that are best diagnoses on conventional X-rays. Early diagnosis of the condition can help increase the patient’s survival rate as specific enzyme replacement therapy has been developed as a potential treatment modality for some patients with MPS-I.



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