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MUCOPOLYSACCHARIDOSIS IN PAEDIATRICS – A CASE REPORT

Radiodiagnosis

Dr. Akshay Magadum* Third Year Resident, Department Of Radio-diagnosis, Smt. B'K Shah Medical Institute & Research Centre, Sumandeep Vidyapeeth Deemed To Be University, Waghodia Vadodara. *Corresponding Author

Dr. Rajesh Rathore Professor , Department Of Radio-diagnosis, Smt. B'K Shah Medical Institute & Research Centre, Sumandeep'vidyapeeth'deemed To Be University, Waghodia Vadodara.

ABSTRACT

Mucopolysaccharidoses (MPS) are a group of inherited lysosomal storage disorders, having in common an excessive accumulation of mucopolysaccharides secondary to deficiencies in specific enzymes (lysosomal hydrolases) responsible for their degradation which leads to accumulation of the GAGs, dermatan sulfate and heparan sulphate. The spectrum constitutes a disorder with severe involvement and CNS disease Hurler disease (HPS I H), a chronic disease without CNS disease Scheie disease (HPS I S5) and the intermediate Hurler/Scheie disease(HPS I HIS). The skeletal disease dysostosis multiplex (DM) is observed in severe variants of MPS I which is of high risk. MPS IH (Hurler Disease) affected children show a spinal 'gibbus' deformity, persistent nasal discharge, middle ear infections and recurrent upper respiratory tract infections.

KEYWORDS

Mucopolysaccharidosis, X-ray, Paediatrics

INTRODUCTION

The mucopolysaccharidoses constitute a group of hereditary disorders having in common an abnormality in glycoprotein or mucopolysaccharides metabolism secondary to deficiencies in specific enzymes leading to excessive accumulation of glycosaminoglycans. The urine Glycosaminogycans are detected by immunoassay which is gold standard.

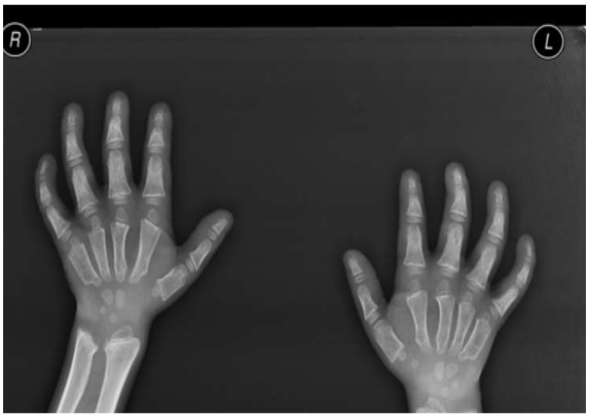
MPS are classified into MPS I-H (Hurler's syndrome), MPS II (Hunter's syndrome), MPS III (Sanfilippo's syndrome), MPS IV (Morquio-Brailsford syndrome), MPS I-S (Scheie's syndrome) and MPS VI (Maroteaux- Lamy syndrome). The Hunter type of MPS is inherited as an X-linked recessive; the others are autosomal recessive in nature.

CASE REPORT

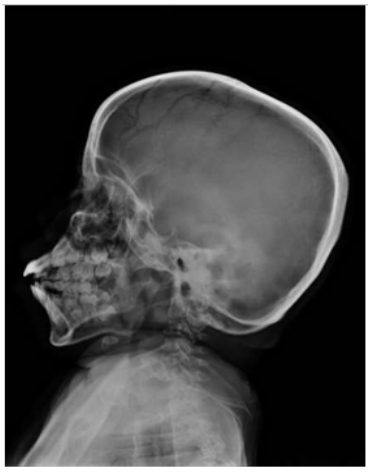
A 5 year old male child with mental retardation was brought by his parents to the hospital for the treatment of recurrent ear discharge and hearing loss. He had coarse facial features, macrocephaly, hepatosplenomegaly, stiffness in multiple joints and had progressive vision loss.

RADIOLOGICAL PRESENTATION

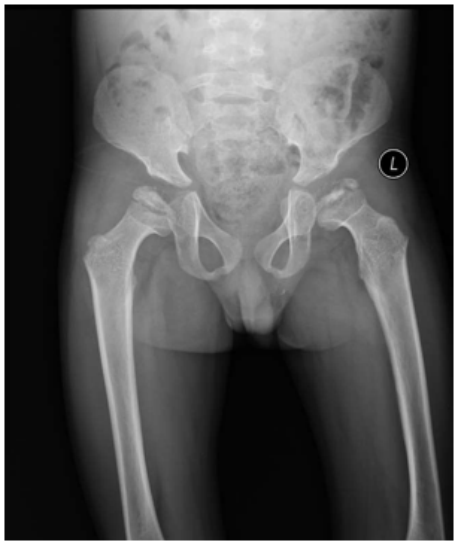
- The skull appears large(macrocephaly) with thickened calvarium.
- Kyphosis may be present.
- The pelvis is usually poorly formed.
- The clavicles are thickened, irregular and shorter.
- The ribs are oar-shaped.
- The phalanges are short, broad and trapezoid with widening of the diaphyses.



X-ray Both Hand : Shows Broad Phalanges With Tapering Of Proximal Ends Of The Metacarpals



X-ray Skull Lateral View: Shows Macrocephaly With Frontal Bossing With Calvarial Thickening. The Sella Is Enlarged And Appears J- Shaped. The Facial Bones Appear Smaller Than The Calvarium. Dental Malformation Is Also Noted.



X-ray Pelvis With Both Hips A.p View: Shows Widely Flared Iliac Wings With Inferior Tapering. Maldeveloped And Fragmented Femoral Epiphyses Are Notewith Associated Metaphyseal Irregularity And A Shallow Acetabulum. Coxa Valgus Deformity Is Also Noted.

MANAGEMENT OF THE CONDITION

- Joint stiffness: exercises at home, yoga and physiotherapy.
- Hearing loss: control of infection and hearing aids like BAHA.
- Hydrocephalus: Ventriculo-Peritoneal shunt.
- Corneal clouding: Corneal transplantation
- Cardiovascular disease: Valve replacement- Mitral and aortic valves are affected the most.
- Obstructive airway disease: Tracheostomy has good success rates in patients having sleep apnea.

Treatment

- ENZYME REPLACEMENT THERAPY :-
- Laronidase is the synthetic form of human enzyme alpha- L-iduronidase produced by recombinant DNA technology.
- It is indicated to treat MPS type I.
- Idursulfase is purified form of human iduronate-2-sulfatase, a lysosomal enzyme used in treatment of MPS I.
- Bone marrow transplantation (BMT):

Bone marrow transplantation (BMT) has shown better results in the treatment of MPS conditions, especially Hurler syndrome. Children treated with BMT have an increased lifespan compared to those untreated children. Untreated children commonly died of cardiorespiratory conditions in the first decade of life.

CONCLUSION

- Severe form of the disorder shows dysostosis multiplex, which refers to multiple skeletal abnormalities seen on x-ray.
- Early diagnosis is important for identification of the condition and initiation of treatment.
- Different types of MPS are diagnosed using enzyme assay techniques but radiography too plays a very important role in diagnosis of the condition and differentiate the two major types of MPS namely Hurler's syndrome and Morquio's syndrome.

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