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Case Report

Case Report of Mucopolysaccharidosis type -1 (MPS-1)

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ABSTRACT

Mucopolysaccharidosis (MPS) represents a heterogenous group of inheritable lysosomal storage disease in which accumulation of undegraded glycosaminoglycans (Gags)- {dermatan sulfate & heparan sulfate}interfere with cell functions and leads to progressive damage of affected tissue. Radiological features; skeletal radiograph and MRI allows theassessmentof the severity of the disease (10). The case highlights that abnormal clinical symptoms, such as growth failure, coarse facial features, and joint problems, are key points for further investigation relating to mucopolysaccharidosis disease. Deficiency of α-L-iduronidase results in a wide range of clinical involvement from severe Hurler disease to mild Scheie disease (MPS I S) & intermediate form Hurler-Scheie disease (MPS I HS) (2).

Key Words: *Mucopolysaccharidosis, Hurler syndrome, Scheiedisease, Glycosaminoglycans, skeletal radiograph*

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INTRODUCTION

The mucopolysaccharidoses (MPS) are described as heritable lysosomal storage disorders which show an Autosomal recessive inheritance pattern and are caused by a deficiency of glycosaminoglycan (GAG) which is a degrading enzyme.MPS-1- Hurler syndrome occurs due to mutation in the IDUA gene present on chromosome 4p16.3 which is encodingα-Liduronidase. Accumulation of GAGs—like (dermatansulfate, heparansulfate, keratansulfate, and chondroitin sulfate)—in lysosomes causes progressive damage to affected tissues, including the heart, respiratory system, bones, and central nervous system, etc. Three major clinical subgroups are described among the mucopolysaccharidosis type 1: Hurler (MPS-IH), Hurler-Scheie, and Scheie. Musculoskeletal manifestations are common features of all types of mucopolysaccharidosis and can be predominant among attenuated phenotypes (4, 10, 2,).

CASE REPORT

A 1-year-old female child first in birth order came to the pediatric department with acomplaint of fever, cough with expectoration, and Increase head size since birth. There was no significant perinatal history. On examination coarse facies, clouding of the cornea, broad forehead, depressed nasal bridge, and gibbous were seen. The anterior fontanelle was open and the head circumference was 51cm.on auscultation murmurs were heard. During per abdomen examination, the liver could be felt 4 cm below the subcoastal margin-suggestive of hepatomegaly. Clawed fingers of both hands of the patient. Excessive hair growth, gibbous deformity, and a hyper pigmented patch were also noted in the lumbosacral region. An enlarged abdomen and umbilicalhernia were also noted. All baseline investigations were done, and CBC was suggestive of low Hb microcytic hypochromic anemia and hypothyroidism ESR and ammonia were raised. USG's whole abdomen was done which suggests mild hepatomegaly. A 2D echo was done and was suggestive of the thickened aortic valve, mild mitral regurgitation, and mild mitral valve prolapse.

Radiographic findings of dysostosis multiplex cover virtually the entire skeleton. X-ray hands show widening of phalanges and metacarpals with proximal tapering, bullet-shaped phalanges. X-ray hand revealed a delayed appearance of carpal bones with short and broad metaphysic & proximal narrowing of the metaphysis, hypoplasia of left ulna with metaphyseal irregularity. X-ray forearm showed widened diaphysis in both the radius and ulna. X-ray spine revealed anterior beaking of vertebral bodies. X-ray chest showed broad oar-shaped ribs. MRI brain on T2 sequence ventricular system and cortical sulci are slightly prominent. urine mucopolysaccharidoses screening was done and it turned out to be positive.

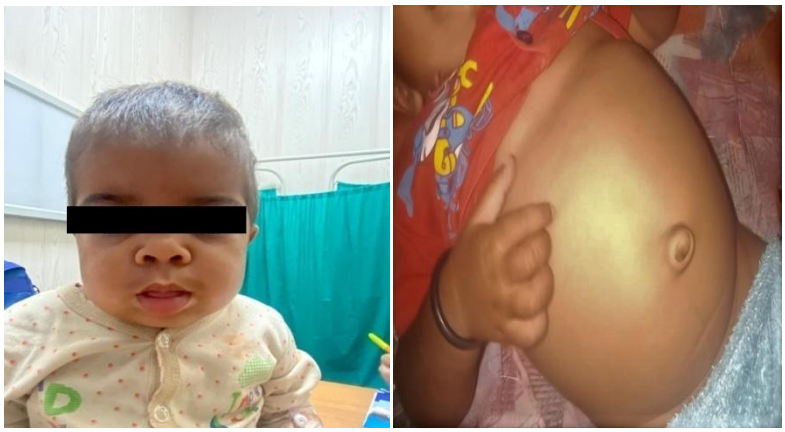

Fig1: Coarse facial features, broad forehead, **Fig2**: Abdominal distention and depressed nasal bridge, Macroglossia, and umbilical hernia were noted gingival hypertrophy was reported in this case. In this case.

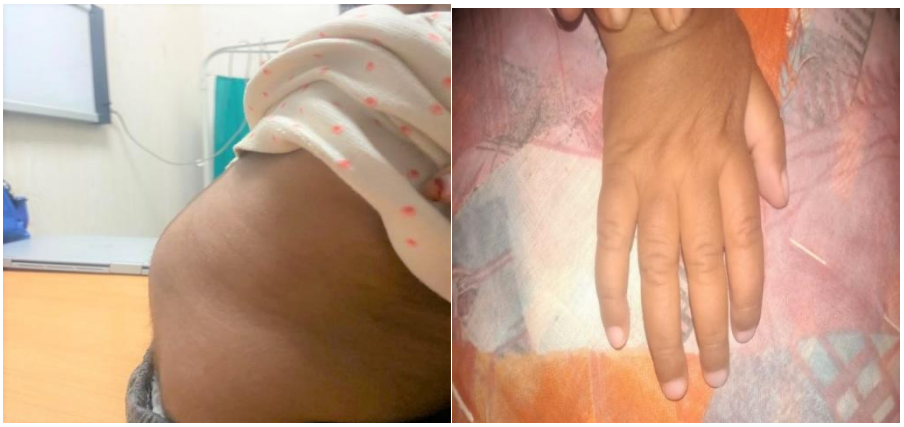

FIG3: Shows gibbus deformity, **FIG 4:** Mild claw deformity in hands, increased hair growth.

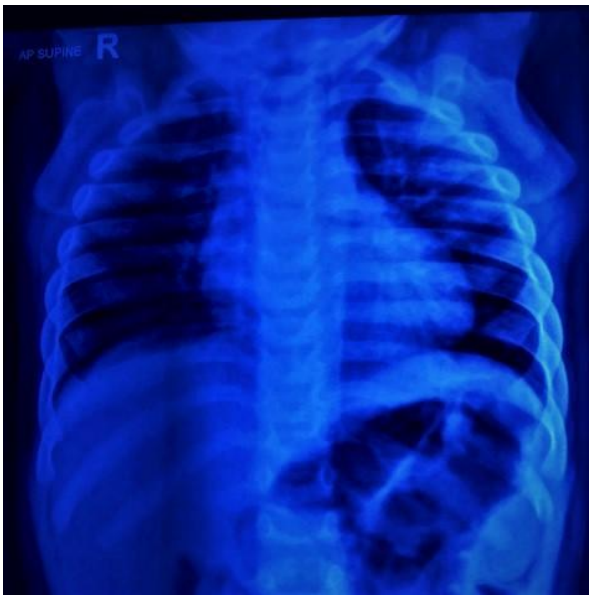

Fig 5:-Chest Radiography P.A view: Paddle-shaped widenedribs on chest X-ray Oar-shaped ribs with narrowing at the vertebral ends and broadening at the sternal ends of the patient included in the case report. Mild Cardiomegaly is also seen. The mild thick and irregular clavicle bone is noted.

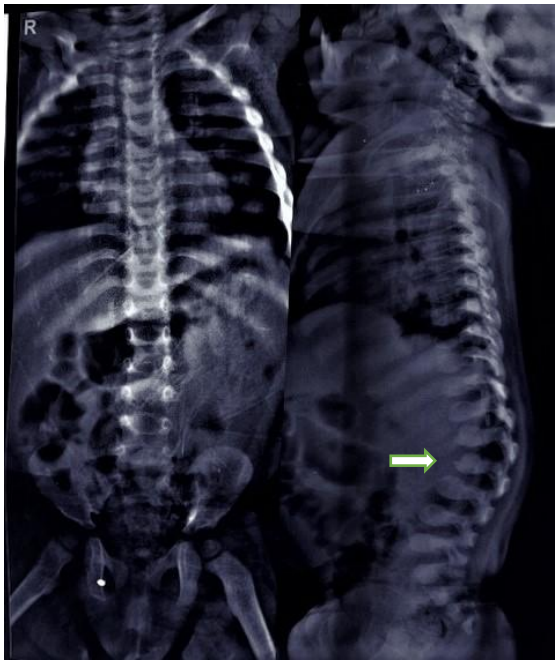

Fig 6: **X-ray Dorsolumbar spine (Anteroposterior and Lateral view**) reveals Anterior Beaking of vertebral bodies. Oar-shaped ribs with narrowing at the vertebral ends and broadening at the sternal ends of the patient and mild metaphyseal irregularity are seen in both femurs with diaphyseal widening included in the case report.

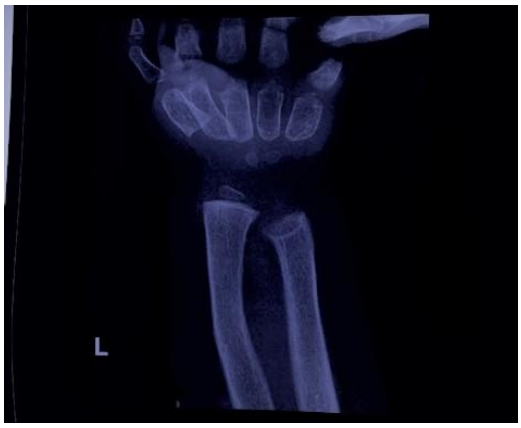

Fig 7: -X-ray forearm with hand- AP radiograph of hand and wrist shows widened metaphyses and diaphyses with bullet-shaped phalanges with the proximal pointing of the second to fifth metacarpals of the patient included in the case report.

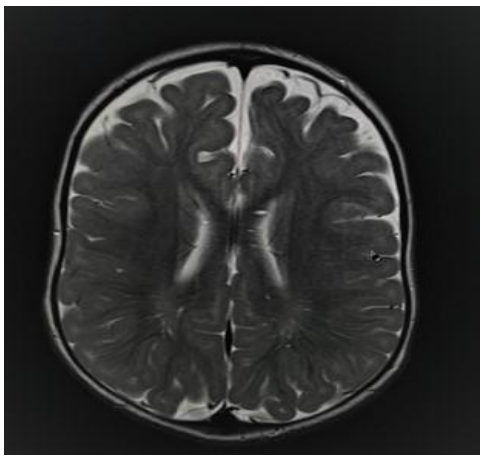

FIG 8: MRI brain showing mildprominence of frontal convexity sulci and ventricular system also appears to be prominent.

DISCUSSION

Mucopolysaccharidoses are a group of autosomal recessive inheritance disorder And the absence of Enzyme Alpha-L-iduronidase results in the accumulation of glycosaminoglycansIn lysosomes of various tissues of the body, resulting in organ damage and causing mental retardation, stunted growth, skeletal malformations, stiff joints, corneal clouding, thick lips, macroglossia with spaced and hypoplastic teeth, and excessive excretion of the GAG's (heparan sulfate and dermatan sulfate) in the urine, as observed in our case. The other manifestations are organomegaly, mental retardation, and umbilical hernia. The Urine Cetyl Trimethyl ammonium bromide (C-TAB) test is the most important qualitative test for screening for MPS. MPS-1 can also present with various other features like - variable degree of growth failure, intellectual disability, coarse facial features, joint contractures, corneal clouding, and, dysostosis multiple, Children with Hurler syndrome also have a loss of vision (due to pigmentary retinopathy), hearing loss, otitis media, obstructive sleep apnea, and recurrent respiratory infections. Cardiac involvement results in regurgitation across valves, cardiomyopathy, and arrhythmias.

Radiographical examination (Fig 5, 6, 7, 8) revealed characteristic skeletal dysplasia (Dysostosis multiplex). X-ray hand revealed a delayed appearance of carpal bones with short and broad metaphysic & proximal narrowing of the metaphysis. X-ray forearm showed widened diaphysis in both the radius and ulna. X-ray spine revealed anterior beaking of vertebral bodies X-ray chest showed broad oar-shaped ribs. X-ray pelvis revealed narrowing of the lower part of the pelvis with shallow acetabular fossa. X-ray of lower limbs showed widened diaphysis of both femurs with a coarse trabecular pattern.

Early diagnosis is the key to the prompt treatment of such cases of inborn metabolic errors. Most children with hurlers appear normal in the initial stages, but if left untreated, the disease can be progressive and may lead to several complications. Allogeneic hematopoietic stem cell transplantation (HSCT) along with Enzyme replacement therapy (ERT) with human recombinantlaronidase (peritransplantation) is currently the gold standard and is treated before 2–2.5 years of age.

CONCLUSION

Characteristic coarse facies, other clinical features, and Radiological features suggest Dysostosis Multiplexas described above in our case. Urine examination screening for mucopolysaccharidosis was positive. EXOME genomic sequence for the same was also sent. Therefore concluding all the findings they are in favor of MPS 1 i.e Hurlers disease. MPS I-H is a rare, life-threatening multisystem disorder. It is the most frequent type of MPS reported. Early detection of the disease and appropriate management through a multidisciplinary approach is recommended to improve the quality of life.

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Patient consent – Due consent from the patient's parents was taken for the publication and research work.
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