

## Hurler's Syndrome: A Rare Case Report

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**Abstract:** Hurler's syndrome is a rare lysosomal storage disorder caused by deficiency of lysosomal enzyme  $\alpha$ -iduronidase and has an autosomal recessive inheritance. Mucopolysaccharidosis-1(MPS1) is classified into mild (Scheie syndrome), intermediate (Hurler- Scheie syndrome), severe (Hurler syndrome). We are reporting a case of Hurler's syndrome which affected a male child of 13 years. The child has  $\alpha$ -iduronidase enzyme deficiency which in turn leads to deposition of dermatan and heparan sulphate in multiple tissues leading to progressive deterioration.

**Keywords:** Hurler's syndrome, Lysosomal enzymes,  $\alpha$ -iduronidase, Mucopolysaccharidosis-1(MPS1), Dermatan, Heparan sulphate.

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### INTRODUCTION

Hurler's syndrome is a rare lysosomal storage disorder caused by deficiency of lysosomal enzyme  $\alpha$ -iduronidase and has an autosomal recessive inheritance which results in progressive accumulation of glycosaminoglycans (GAG) within the lysosomes, later leading to multi-organ dysfunction and damage. MPS1 individuals are unable to degrade the GAG, dermatan and heparan sulphate, which provide structural support to extra cellular matrix, cartilaginous tissues like heart valves and joints [1]. MPS1 has a prevalence of 1 in 1,00,000 of live births [2] and 20% of which represented by Intermediate type (Hurler- Scheie syndrome). We report a case of Hurler's syndrome in this article.

### CASE REPORT

A 13 year male child presented with breathlessness of grade 3, distention of abdomen, edema feet and stunted growth. On general examination short stature (Fig. 1), bossing of forehead, depressed nose, clouding of cornea, coarse facies (Fig. 2), short neck, low set ears (Fig. 3), ascites, umbilical hernia, thick short palms and soles (Fig. 4), pulse 100/minute with regular rhythm, BP 100/70 mmHg, temperature normal, respiratory rate 20/minute. His cardiac examination revealed elevated JVP, apex shifted down and out in 6<sup>th</sup> space, p2 loud, no murmurs, bilateral crepitations in lungs and hepatosplenomegaly suggestive of cardiomyopathy and pulmonary arterial hypertension with heart failure. On investigation ECG revealed P pulmonale, RVH and Right ventricular overload (Fig. 5). Echocardiogram showed enlargement of both ventricles with ejection fraction of 35%.

Radiological findings include cardiomegaly and oar or paddle shaped ribs on x ray chest (Fig. 6), beaking of inferior margins of vertebrae on spine x-ray (Fig. 7), widening of femoral neck and winged pelvic bones on x- ray pelvis (Fig. 8) and bullet shaped phalanges, widening of metacarpals on hand x- ray (Fig. 9). All these features are suggestive of Hurler's syndrome. Patient is kept on diuretics, ACE inhibitors, antibiotic to treat his CHF and secondary chest infection.



**Fig. 1: Short stature**



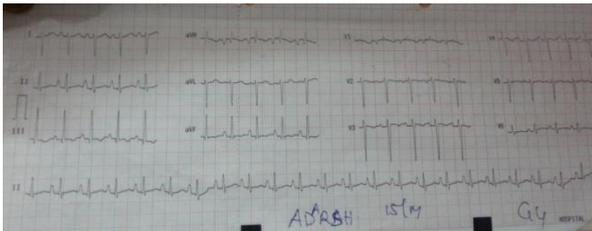
**Fig. 2:** Coarse facies, cloudy cornea, depressed nose, bossing of forehead



**Fig. 3:** short neck, low set ears



**Fig. 4:** Ascites, umbilical hernia, thick short hands



**Fig. 5:** ECG showing P-pulmonale, RVH



**Fig. 6:** Cardiomegaly, oar shaped ribs



**Fig. 7:** inferior beaking of vertebrae



**Fig. 8:** Winged pelvic bones and widening of femoral neck



**Fig. 9: bullet shaped phalanges, widening of metacarpals**

## DISCUSSION

Hurler's syndrome, caused by deficiency of the lysosomal enzyme,  $\alpha$  1-iduronidase, is traced to the chromosome 4p16.3 [3]. The clinical manifestations include short stature, mental retardation, enlarged skull, low nasal bridge, thick coarse facies, deafness, short hands and feet with vertebral anomalies like atlantoaxial subluxations [4], inferior beaking of vertebral bodies and infiltration of duramater and cervical cord with Mucopolysaccharides [5]. Storage disorders that produce skeletal abnormalities are collectively termed as "dysostosis multiplex." Cardiovascular manifestations like cardiomyopathy, pulmonary arterial hypertension and heart failure are commonly noted in MPS1. Frequent upper and lower respiratory tract infections are commonly encountered secondary to enlarged tonsils, adenoids. Most often the affected children usually die within 1<sup>st</sup> decade due to either cardiac or respiratory failure. The diagnosis of MPS1 is made by finding out lysosomal enzyme,  $\alpha$ -iduronidase, levels in plasma, peripheral leucocytes and cultured fibro blasts. In our case, most of the findings classically described in literature are found out. Life expectancy can be enhanced by enzyme replacement with Laronidase and bone marrow transplantation [6]. When there is positive family history of Mucopolysaccharidosis, genetic counselling and testing for the enzyme should be recommended for the newlywed families.

## CONCLUSION

In this case report, we have come to the diagnosis of Hurler's Syndrome, Mucopolysaccharidosis type 1 by the clinical manifestations and radiological evidence of specific features.

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