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Case Report

# An Interesting Case of Facial Dysmorphism with Hepatosplenomegaly

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## Abstract

Mucopolysaccharidosis (MPS) is a rare lysosomal storage disorder with an incidence of 1 in 2 lakhs. It is characterised by deposition of GAGs in various organs due to specific lysosomal enzyme deficiency. Early diagnosis and multidisciplinary approach is vital in preventing mortality and morbidity. If left untreated, death can result by 10 years due to cardiorespiratory failure. Discouraging consanguineous marriage and genetic counselling is important in reducing the incidence of the disease. Here, we report the case of a 5 year old child who presented with growth retardation.

**Keywords:** GAG (Glycosaminoglycans); Hurler's Syndrome; MPS-1 (Mucopolysaccharidosis Type-1); Multidisciplinary Treatment (MDT)

## Introduction

Mucopolysaccharidoses are a group of metabolic disorders caused by the absence or malfunctioning of lysosomal enzymes needed to breakdown glycosaminoglycans that leads to accumulation of GAGs in cells, blood and connective tissues resulting in progressive cellular damage affecting appearance, physical abilities, organ and system functioning.

## Case Report

A 5 year old boy, was brought to the outpatient department with complaints of poor vision, stunting and delay in achievement of milestones in the form of inability to walk steadily, feed oneself, peer play, talking clearly (utters few incomprehensible sounds). No h/o fever, chronic cough, feeding difficulty, constipation, difficulty in breathing, failure to thrive, abnormal movements. History of respiratory ailments treated on OPD basis multiple times in the past present. He was born by vaginal delivery. Antenatal and postnatal periods were uneventful. No history of tuberculosis, past surgeries or long term medication. Child was born out of first degree consanguineous marriage to a couple in late twenties (elder sibling; who was 9 year old was apparently normal). He was on full family diet with no calorie or protein deficit. On examination vitals were stable. Anthropometry showed stunting grade III with upper segment and lower segment ratio of 1.6. Height age-2 years, weight age-5 years, bone age-3 years. Head circumference > 3SD (56 cm) Head to toe examination revealed facial dysmorphism with macrocephaly, frontal bossing, hypertelorism, corneal clouding, broad nasal bridge, prominent supraorbital rim, widely spaced teeth, short stubby hands, kyphoscoliosis were present. Respiratory system examination showed conducted sounds present with normal breath sounds. Ejection systolic murmur was heard at the aortic area on precordial auscultation. Per abdomen examination showed

moderate hepatosplenomegaly. He had no focal neurological deficits. Based on history and examination, preliminary diagnosis of Mucopolysaccharidosis was suspected and a radiological survey was done which showed trigger digits and kyphoscoliosis at T6-L2 level with ovoid vertebral bodies. Screening with urinary excretion of GAGs was positive. Diagnosis was confirmed by doing enzyme assay of alpha L iduronidase in the peripheral blood leukocyte. He was managed with multidisciplinary approach involving Paediatrician, Endocrinologist, Orthopedician, Dentist, Cardiologist, Neurologist, Developmental Pediatrician, Gastroenterologist and Ophthalmologist. Child was referred to higher centre for enzyme replacement therapy.

## Discussion

Hurler's syndrome (MPS-type1) is an autosomal recessive disorder caused by mutations of the IDUA gene on chromosome 4p16.3 encoding alpha-L-iduronidase. Affected infants appear normal at birth, but inguinal hernia is usually present. Majority have recurrent upper respiratory tract infections. Valvular heart disease involving mitral and aortic valves, narrowing of coronaries, acute cardiomyopathy can occur. Obstructive airway disease, notably during sleep, may necessitate tracheostomy. Language skills are poor due to combined conductive and neurosensory deafness, and macroglossia. Progressive ventricular enlargement with increased intracranial pressure caused by communicating hydrocephalus can also occur. Corneal clouding, glaucoma, and retinal degeneration are common. Radiographs shows dysostosis multiplex. The earliest sign being thick ribs and ovoid vertebral bodies. Obstructive airway disease, respiratory infections, and cardiac complications are the common causes of death [1,5]. Diagnosis is based on clinical suspicion, radiological survey, urinary GAG screening tests and confirmation is with enzyme assay. Prenatal diagnosis and neonatal blood spot screening help in early diagnosis [1,4]. Treatment include enzyme replacement, haematopoeitic stem cell transplant therapy, substrate reduction by flavonoids, gene silencing [1,3], bone marrow transplantation and cord blood transplantation (before 24 months) [1,2]. Symptomatic management with Ventriculoperitoneal shunting, behavioral medication, corneal transplant, hearing aids, adenotonsillectomy, CPAC at night, endocarditis prevention, dental care, corrective osteotomies, physical therapy are important for better quality of life [1].

## Conclusion

MPS is a rare genetic disorder. In a child with growth delay, facial dysmorphism and hepatosplenomegaly it should be borne in mind as one of the differential diagnosis. With timely diagnosis and appropriate management involving multidisciplinary approach, good quality of life can be assured. Genetic counselling and prenatal diagnosis is important to reduce the occurrence of the disease.

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**Volume 2 Issue 8 August 2021**