HURLER’S SYNDROME- A RARE CASE REPORT

G. Jayanthi¹, K. Somvya², Dr G. Ramya Bala Prabha Naidu, Sushanta Kumar Das and Dr K. Abbulu

CMR College of Pharmacy, Kandlakoya, Hyderabad, Telangana- 501401.

*Corresponding Author: G. Jayanthi
CMR College of Pharmacy, Kandlakoya, Hyderabad, Telangana- 501401.

ABSTRACT
Mucopolysaccharidosis type I (MPS I H, Hurler syndrome) is a rare autosomal recessive inborn deficiency in the metabolism of glycosaminoglycans (GAGs) heparan sulfate and dermatan sulfate, resulting from deficiency of Alpha-L-iduronidase enzyme. This condition is characterized by accumulation of incompletely degraded glycosaminoglycans into various organs of body, which leads to impairment of organs and body functions. Such children appear nearly normal at birth; however, if left untreated, show a progressive mental and physical deterioration leading to death due to cardiorespiratory failure before the second decade of life.

KEYWORDS: glycosaminoglycans, Hurler syndrome, Mucopolysaccharidosis type I, Autosomal recessive, iduronidase.

INTRODUCTION
Mucopolysaccharidoses are an inborn heterogeneous group of rare metabolic disorders inherent as autosomal recessive traits, due to deficiency or absence of lysosomal hydrolase - iduronidase enzyme activity. The defect has been mapped to the chromosome band 4p16.3,[1,2] Hurler syndrome (MPS I - H) is the most common and severe form of mucopolysaccharidoses.[3] Deficiency of this enzyme results into a wide range of phenotypes including Hurler’s (severe), Scheie’s (mild) and Hurler-Scheie (intermediate) syndromes.[3] MPS I has an estimated incidence of 1 case per 100,000 live births[6,7] and the attenuated type represented about 20% of the total MPS I population.[4]

Mucopolysaccharidosis I (MPS I) is a rare inherited disorder that belongs to a group of clinically progressive disorders and is caused by the deficiency of the lysosomal enzyme, α₁-iduronidase. MPS I has been recently classified into a severe (Hurler syndrome) and an attenuated type (Hurler-Scheie and Scheie syndromes). The purpose of this was to describe a rare case of Mucopolysaccharidosis type I (MPS I H, Hurler syndrome) affecting a 3-year-old child.

CASE REPORT
A 3 year old male child was reported to pediatrics with chief complaints of vomiting since 10 days immediately after feeds and unable to see food and people placed in front of him; H/o of trauma 15 days ago where child allegedly fell from 1st floor of house playing by himself . On examination child has coarse facial features like hat nasal bridge which is prominent on forehead.(Figure 1) macrocephaly, corneal clouding (Figure 2), swelling over lumbar spine, monogloid spots, lower seat ears.

INVESTIGATIONS
- CT scan revealed synotosis of saggital suture; Scaphocephaly noted.
- X ray report shows that
  - Skull: J shaped AP diameter more than the width.
  - Hands: pointing of proximal metacarpals were noted like Mucopolysaccharidosis (Hurler’s syndrome).
  - Pelvis: Flaring of iliac wings with shallow acetabulum noted.
  - Chest: Normal heart and lungs clear. Broad anterior ribs.
  - Ultrasound of abdomen: Normal
  - Patient was advised with urine GAG TESTING.

DISCUSSION
Enzyme Alpha-L-iduronidase is responsible for the degradation of the glycosaminoglycans (GAGs), and its absence results into accumulation of heparan sulfate and dermatan sulfate in lysosomes of various tissues of the body, resulting in organ damage,[2,3] and causing mental retardation, stunted growth, skeletal malformations, stiff joints, corneal clouding, effect on cardiorespiratory system, thick lips, macroglossia with spaced and hypoplastic teeth, and excessive excretion of the heparan sulfate and dermatan sulfate in the urine.

In this case child is effected with coarse facial features like hat nasal bridge which is prominent on forehead (Figure 1), macrocephaly, corneal clouding (Figure 2), swelling over lumbar spine, monogloid spots, lower seat ears.
Children with Hurler's syndrome appear nearly normal at birth; it is considered to be incurable; however, as multiple organs are involved a multidisciplinary approach is needed to sustain and improve the quality of life. There are currently two different well-established approaches for the treatment of Hurler syndrome if diagnosed early, and these includes Hematopoietic stem cell transplantation (HSCT) and enzyme replacement therapy with alpha-L iduronidase enzyme to stabilize or reverse many aspects of Hurler syndrome.  

CONCLUSION
There exists a need for prevention, early diagnosis and management of Hurler Syndrome through a multidisciplinary approach to improve the quality of life. With the advent of hematopoietic stem cell transplantation and, more recently, enzyme replacement therapy, there exists a need for early diagnosis, better disease recognition and management. Early diagnosis is crucial for the best therapeutic outcomes with both ERT and HSCT.

REFERENCES