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Case Report

Hunter's syndrome: a rare case report

Akhila, Anupama Singh, Jayant Raj, Amit Shekharay, Vinod K. Mishra, Monika Singh, Om P. Singh*

Department of Pediatrics, NMCH, Jamuhar, Sasaram, Rohtas, Bihar, India

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***Correspondence:**
Dr. Om P. Singh,
E-mail: drops0105@gmail.com

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ABSTRACT

Hunter's syndrome also known as mucopolysaccridosis-II (MPS-II) is a very rare X-linked recessive disease. It is a chronic progressive metabolic disease. The incidence of this condition is 1 in 1, 62,000 live births, mostly males. There is excessive accumulation of glycosaminoglycan (GAGs) in the lysosome in various parts of body due to the deficiency of enzyme iduronate sulphatase (IDS) and manifests with variety of clinical signs and symptoms. Four novel mutations in IDS gene are reported among families of Hunter syndrome tested by Sanger sequencing of IDS gene.

Keywords: Mucopolysaccharidosis, Glycosaminoglycan, Iduronate sulphatase, Iduronate sulfatase gene, Lysosomal storage disorder

INTRODUCTION

Mucopolysaccridosis-II (MPS-II) is an X-linked multisystem disorder due to deficiency of an enzyme iduronate sulphatase (IDS).[1] Mutation in IDS gene is responsible for this disease.[2] This enzyme IDS is responsible for breakdown or metabolism of various substances including glycosaminoglycan (GAGs). These GAGs build up in lysosomes leading to progressive damage and dysfunction in cells, tissues and organs throughout the body. This results in wide variety of signs and symptoms in Hunter's syndrome. The incidence of the disease is 1 in 100,000 to 150,000 male births.[2,3]

CASE REPORT

This 11-year-old male child presented to us at the department of pediatrics at tertiary care teaching center in Sasaram, Bihar for enzyme replacement therapy. He was first diagnosed at AIIMS Delhi four years back. The child had inability to speak, hyperactivity, coarse facial features in the form of broad nose with prominent flared nostrils along with broad thick lip, enlarged jaw and hand, along with early appearance of secondary sexual characters, Tanner's staging four (Figures 1 and 2).

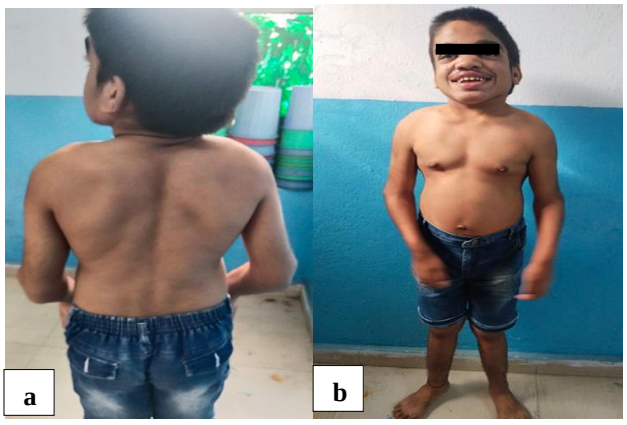

Figure 1 (a and b): Hunter's syndrome with scoliosis, typical facies, large head and secondary sexual characters.

Child's vitals were stable. Anthropometrically, height was 136 cm, head circumference 62 cm, arm span 128 cm, US:

LS ratio was 0.3. The child had, gum hyperplasia, thick and large tongue, frontal bossing, short neck, varus deformity of upper limbs. He also had spinal deformities in the form of scoliosis. He had enlarged abdomen due to hepato-splenomegaly along with various dental anomalies. Additionally, he had mixed sensory-neural hearing loss. Investigations revealed urine glycosaminoglycan level 271.6 mg /mmol. ABR test report inferred B/L sensori-neural hearing loss, 2D echo showed mild AR. The child is under continuous care with occupational and behavioral therapy. Child is receiving regular enzyme replacement therapy from this center.

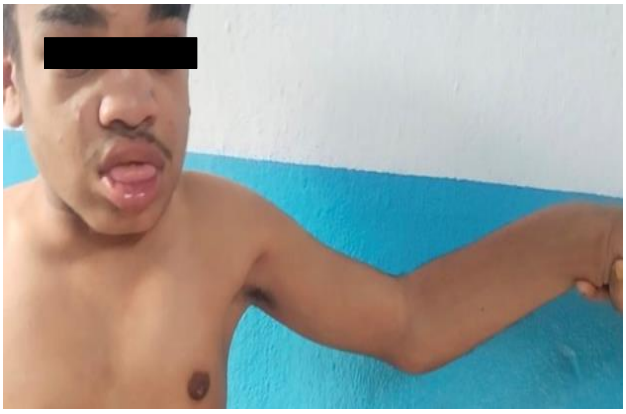

Figure 2: Image showing upper limb deformity, thick lips, broad nose and coarse facial features.

DISCUSSION

Hunter's syndrome is a rare X linked inherited disorder which occurs due to deficiency of lysosomal hydrolase Iduronate 2-sulphatase which helps in degradation of heparan and dermatan sulphate leading to accumulation and dysfunction of multiple organ systems.[1,4] It represents a severe clinical phenotype. Hunter's syndrome is a slowly progressive disease and is difficult to diagnose early. Early signs and symptoms emerge by 2 to 4 years and the severity is different between patients with a variable age of onset and variable rate of progression.[1-3] In some cases cognitive impairment may manifest. Earliest visible sign of Hunter's disease is unusual facial features enlarged abdomen tongue and tonsils, notable features are a distended abdomen due to enlarged liver and spleen however their function is normal.[1,4,5] Hernias are commonly seen, umbilical hernia being the commonest. In the respiratory system airway obstruction is a common symptom, recurrent ear infections being the commonest. There may be progressive skeletal changes with stiffening of joints and limitation of movement due to persistent tightening of tissues. In the severe forms there may be neurological manifestations like intellectual impairment, behavioral problems, cognitive impairment, hyperactivity, ADHD, autism, OCD along with general aggressiveness.[5,6] Treatment of these children involves a multidisciplinary approach involving a primary care pediatrician, neurologist, ENT specialist, orthopedician, dental surgeon and physiotherapist.[3,7] The aim of treating these children is to replenish the deficit enzyme and provide symptomatic therapy. Chinawa et al did a urine chemistry which showed normal urinary mucopolysaccharide level Ogunbiyi et al made the diagnosis mainly based on physical and radiological features, while in addition to these parameters.[6,7] Enzyme replacement therapy has been documented to be resulting in improvement of joint mobility and respiratory tract infections. The typical clinical features and increased levels of urinary GAGs are diagnostic of MPS-II (Hunters syndrome).

CONCLUSION

Management includes a team approach, including pediatrician as a primary care physician, cardiologist, neurosurgery, eye specialist, orthopaedicians, otorhinolaryngologist and pulmonologist. Regular physiotherapy, speech therapy, audiology, dentistry and behavioral therapy are required. Counseling of the parents is an important part of the management. In severe cases, death usually occurs by age 15. In attenuated cases, patients may survive into their 50s. This child is receiving enzyme replacement therapy regularly every week and doing well on follow up.

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