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HURLER SYNDROME IN A YOUNG ADULT MALE: A CASE REPORT

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Abstract:

Hurler syndrome is a form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by skeletal abnormalities, cognitive impairment, cardio-respiratory problems, hepatosplenomegaly, and short life span. Patients typically present within the first year of life with developmental delay along with musculoskeletal abnormalities including short stature, dysostosis multiplex (progressive skeletal dysplasia), kyphosis, and characteristic coarse facies with a large head, full cheeks, bulging frontal bones, depressed nasal bridge, anteverted nostrils, and enlarged lips. Associated abnormalities include cardiomyopathies, valvular abnormalities and hearing loss. An interesting case of a 20 years old male patient with characteristic musculoskeletal, neurological, cardiac, respiratory and visual defects consistent with findings of mucopolysaccharidosis type 1 has been presented in this case report.

Key Words: *Hurler syndrome, mucopolysaccharidosis, glycosaminoglycans.*

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INTRODUCTION:

Mucopolysaccharidoses refers to a spectrum of metabolic disorders that stem from enzymatic defects that lead to defective metabolism and storage of glycosaminoglycans dermatan sulfate and heparan sulfate which provide structural support to extra cellular matrix and cartilaginous tissues like heart valves and joints[1]. The incidence of the disease is 1 per 100,000 live births[2]. No particular predisposition for ethnicity and gender has so far been found. In this article we have reported the case of a 20 years old young male patient with Hurler Syndrome.

CASE REPORT:

A 20 years old male patient presented with shortness of breath, lower urinary tract symptoms and pedal edema. He faced difficulty in voiding, urinary retention, sense of incomplete emptying, urinary hesitancy, poor stream and dribbling of urine. On general physical examination, he had stunted growth, short stature, frontal bossing, depressed nasal bridge and bilaterally hazy cornea. He had coarse dysmorphic facies with low set ears and short neck. Abdominal examination revealed ascites, hepatosplenomegaly and small para-umbilical hernia. His hemodynamics showed blood pressure of 100/70 mmHg, regular pulse rate of 105 per minute and respiratory rate of 28 per minute. Patient was afebrile.

His cardiovascular examination revealed normal JVP and a mid-diastolic murmur. His chest auscultation revealed bilateral basal crepitations suggestive of pulmonary congestion. His neurological examination showed severe myopia. Hearing was normal. Motor system examination showed 3/5 power in lower limbs and 4/5 power in upper limbs bilaterally. Reflexes were normal and plantars were bilaterally down going. His higher mental functions were intact.

His hematological and biochemical investigations were normal (table 1). His thyroid profile and vitamin D level was also normal (table 2). His ECG showed sinus tachycardia and P-pulmonale (figure 1). His urine routine examination was normal and urine culture revealed no growth. His ultrasound KUB showed prevoid urinary volume of 225 ml and postvoid 200ml, kidneys were normal in size and there was no evidence of hydronephrosis.

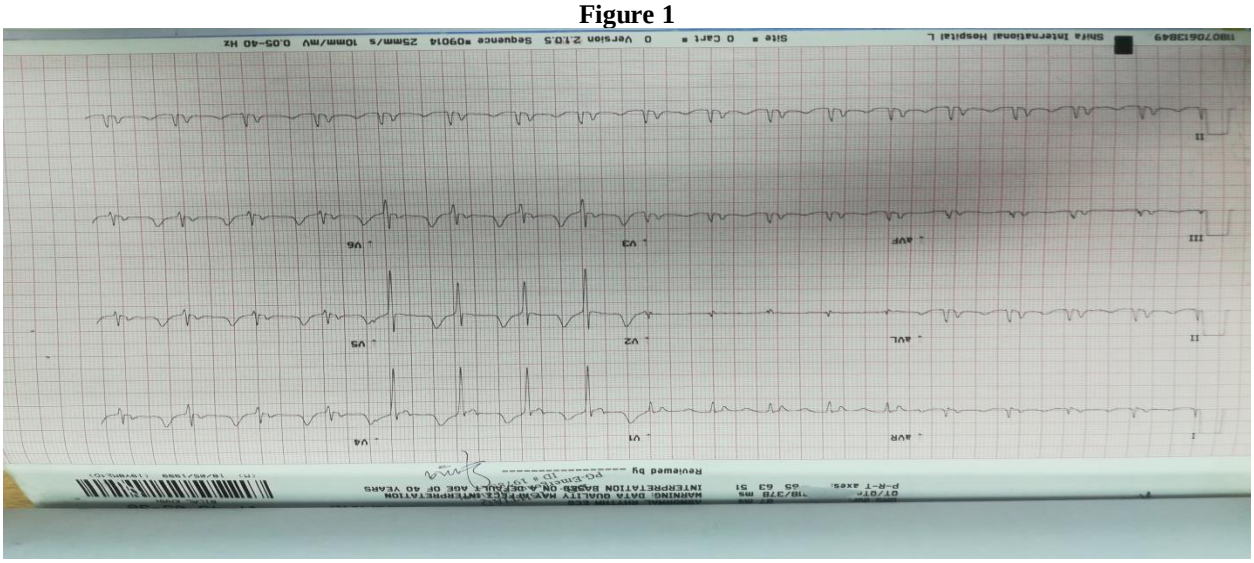
Table 1

WBC, Total	7700/µL
Hemoglobin	13.8 g/dL
Hematocrit	43.6 %
Platelet count	184000/µL
Sodium	142 mEq/L
Potassium	4.4 mEq/L
Bicarbonate	32 mEq/L
BUN	12 mg/dL
Urea	25.68 mg/Dl
Creatinine	0.65 mg/dL
Serum Albumin	3.6 g/dL

Table 2

T3	87 ng/dL
Free-T4	1.32 ng/dL
TSH	5.05 µIU/mL
Intact PTH	85 pg/mL
25-hydroxy Vitamin D	54 ng/mL

Figure 1



His echocardiography showed dilated left atrium, concentric left ventricular hypertrophy, calcified mitral valve with moderate stenosis, mild tricuspid and aortic regurgitation and pulmonary hypertension. Xray findings showed oar shaped ribs, beaked inferior vertebral margins, and widening of phalanges and metacarpals. His detailed MRI lumbar spine report revealed osseus changes in spine and pelvis; scoliosis with reversal of lumbar spine curvature; low lying conus medullaris; mild splitting of cauda equine with a CSF cleft visualized in the midline without any definite osseus or cartilaginous septum. Overall MRI findings were suggestive of a tethered spinal cord.

All the patient's symptoms and findings were consistent with that of mucopolysaccharidosis and a diagnosis of Hurler Syndrome was made. A secondary differential diagnosis of obstructive uropathy with associated pulmonary congestion was made. Patient was started on Tamsulosin which only slightly improved his urinary symptoms. Cystoscopy was planned for the patient which, however, showed no abnormality. The patient was thereafter started on diuretics, ACE inhibitors and antibiotic prophylaxis for chest infections. His follow-up visit showed marked improvement in his symptoms following which his drugs dose was titrated and the regime was continued.

DISCUSSION:

Hurler syndrome is a type of mucopolysaccharidosis I (MPS I) which is typically categorized into three subtypes depending on the severity of symptoms, age of onset and the clinical path of the disease. Scheie syndrome is the mildest form, Hurler-Scheie is the intermediate form, whereas Hurler Syndrome is the most severe form of MPS I[4].

Mucopolysaccharidosis type I (MPS I H, Hurler syndrome) is a rare autosomal recessive defect in the metabolism of glycosaminoglycans (GAGs) heparan sulfate and dermatan sulfate which get deposited in the tissues and are excreted in urine, resulting from deficiency of Alpha-L-iduronidase enzyme[3]. The basic defect has been attributed to chromosome band 4p16.3[5]. This severe inborn error of metabolism leads to early childhood death due to progressive multi-organ failure.

The non-degraded GAGs tend to accumulate within the lysosomes, eventually leading to generalized cellular deterioration. Multi-organ failure ensues. As the number of GAGs increase, the lysosomes eventually rupture and the cells degrade. The detection of GAGs in urine, predominantly heparan and dermatan sulphate, eventually helps confirm the diagnosis[6].

The initial symptoms of the disease which appear in the first few months of life include dysmorphic facies, rhinitis, recurrent upper respiratory tract infections, ascites, hepatosplenomegaly, hernias and kyphoscoliosis of the thoracolumbar spine. The systemic manifestations typically appear after 6 months of age. Progressive psychomotor retardation, impaired hearing, decreased vision, hepatosplenomegaly, severe musculoskeletal manifestations, and cardiovascular and pulmonary failure with death in early childhood are typical features of Hurler Syndrome[4].

Cardiovascular abnormalities like cardiomyopathy, pulmonary arterial hypertension and heart failure are commonly noted in MPS1. Children usually die within the first decade of life due to cardiac or respiratory failure[7]. Confirmational diagnosis of MPS IH is typically made by detecting severely decreased or absent IDUA enzyme activity in peripheral blood leukocytes, filter paper blood spots or fibroblasts, and via genetic analysis[6]. In our case, however, since most of the clinical and musculoskeletal findings cited in the literature were consistent with that of Hurler syndrome, a diagnosis was reached.

There is no definitive treatment for MPS IH. Enzyme replacement with Laronidase enzyme and bone marrow transplantation can help increase life expectancy[6]. In case a positive family history is found, genetic counselling for avoiding consanguineous marriages can be done for prevention of Hurler syndrome.

CONCLUSION:

In this case we reached the diagnosis of hurler syndrome (type 1 mucopolysaccharidosis) on the basis of clinical manifestations and radiological evidence.

REFERENCES:

1. Muenzer J, Wraith JE, Clarke LA. Mucopolysaccharidosis I: management and treatment guidelines. Pediatrics. 2009 Jan 1;123(1):19-29.
2. Hingston EJ, Hunter ML, Hunter B, Drage N. Hurler's syndrome: dental findings in a case treated with bone marrow transplantation in infancy. International journal of paediatric dentistry. 2006 May;16(3):207-12.
3. Matalon R, Dorfman A. Hurler's syndrome: biosynthesis of acid mucopolysaccharides in tissue culture. Proceedings of the National Academy of Sciences of the United States of America. 1966 Oct;56(4):1310.
4. Scriver CR, editor. The metabolic & molecular bases of inherited disease. New York; Montreal: McGraw-Hill; 2001.

5. Banikazemi M. Mucopolysaccharidosis Type I. eMedicine pediatrics: Genetics and metabolic diseases. [Last accessed on 2019, July 2]. Available from: http://emedicine.medscape.com/article/1599374-overview#a0101
6. Aldenhoven M, Boelens J, de Koning TJ. The clinical outcome of Hurler syndrome after stem cell transplantation. Biology of Blood and Marrow Transplantation. 2008 May 1;14(5):485-98.
7. Vaddadi Srinivas D, Ravikumar NA, Srinivasa VR, Vikramvardhan R. Scholars Journal of Medical Case Reports ISSN 2347-6559 (Online).