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Case Report

Hurler syndrome-a case report of infrequently encountered diagnosis

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ABSTRACT

Hurler syndrome also known as mucopolysaccharidosis type 1H (MPS-1H) or gargoylism is an autosomal recessive disorder due to defective gene which encodes for enzyme alpha L-iduronidase (IUDA) located on chromosome 4p16.3 (gene encoding protein iduronidase). In the present case, 4-year Down's syndrome child with coarse facial features, hypothyroidism presented with umbilical hernia. Clinical diagnosis of Hurler syndrome was made corelating with clinical features, X-ray findings.

Keywords: Down syndrome with Hurler syndrome, Autosomal recessive, Umbilical hernia

INTRODUCTION

Hurler syndrome also known as mucopolysaccharidosis type 1H (MPS-1H) formerly known as gargoylism is an autosomal recessive disorder due to defective gene which encodes for enzyme alpha L-iduronidase (IUDA) located on chromosome 4p16.3 (gene encoding protein iduronidase). This enzyme is responsible for breaking down GAGs, deficiency of which results in building of dermatan sulfate and heparan sulfate in several organ like nervous system, skeletal system, eyes and heart.[1] It is classified as lysosomal storage disorder.[2] Overall frequency is one per 1,00,000 male to female ratio is same.[3]

Present case is of 4-year-old male child who presented with hypothyroidism, umbilical hernia, and clouding of cornea.

CASE REPORT

A 4-year-old male child having Down's Syndrome presented with hypothyroidism, umbilical hernia, clouding of cornea.

On examination, he had coarse facial features, short stature, short web neck, frontal bossing, macrocephaly, broad based nasal bridge, frog eye type eye features (protruding widely placed eyes), no spine deformity, umbilical hernia (Figure 1).

Umbilical hernia measured around 5x5 cm with bowel loops within the sac. There was absence of inguinal hernia. ECG showed changes of tachycardia. Spine and chest X-ray were normal. Skull X-ray showed frontal bossing (Figure 2). Ultrsonography of abdomen showed presence of mild hepatomegaly. X-ray abdomen showed presence of ascites (Figure 3). X-ray of upper limb also revealed skeletal abnormalities (Figure 4). All blood indices including LFT and renal profile were normal. Serum TSH level was elevated. The child had delayed milestones.

All the symptoms, signs and radiological findings corelated well with the diagnosis of Hurler syndrome. Hence clinical diagnosis of the Hurler syndrome was made in association with the other radiological investigations.

Figure 1: Coarse facial features.

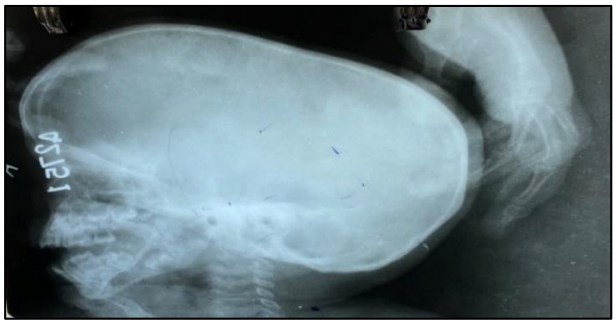

Figure 2: X-ray, frontal bossing.

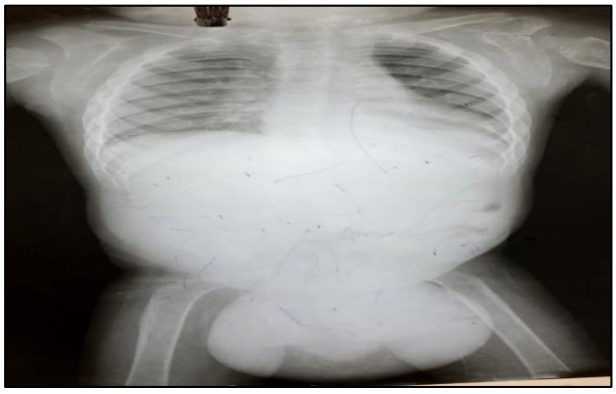

Figure 3: X-ray, ascites, umbilical hernia.

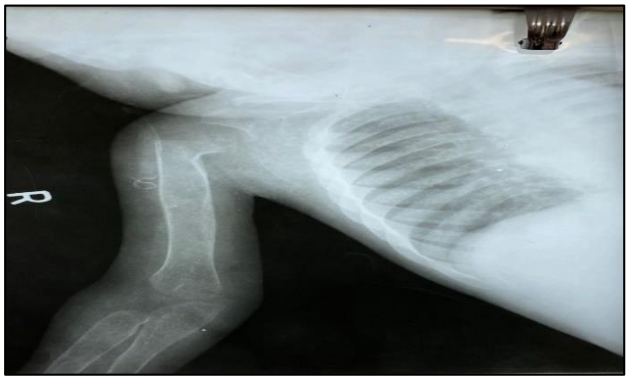

Figure 4: X-ray, skeletal abnormality.

DISCUSSION

Deficiency of lysosomal enzyme IUDA results in accumulation of GAG (dermatan sulfate and heparan sulfate) in the body causing cells to become severely dysfunctional leading to death. there is enlargement and thickening of various organs like the heart, spleen, liver, muscles, connective tissue, joints and central nervous system.[4-6]

MPS1 is subdivided into three subtypes-[7] 1. Hurler syndrome (MPS1H)-most common severe form, average age of mortality within 5 years of life.[3,7] 2. Hurler Scheie syndrome (MPS1H-S) intermediate phenotype, life expectancy into late teens or early twenties. 3. Scheie syndrome (MPS1S)-rare and mild phenotype. Patient die before 25-30 years of age. Clinical diagnosis is made on general appearance-coarse facies (one of the 1st abnormalities to be detected as early as 3-6 months of age), enlarged head, prominent frontal bones, widely placed eye sockets, with protruding eyes, flat nasal bridge with continuous nasal discharge, neck is short. The skull may be elongated, lips may be large, so jaw remains open. Skeletal abnormalities occur about 6 months of age, but clinically seen at 10-14 months of age, may present with debilitating spine and hip deformities, carpal tunnel syndrome, joint stiffness, stop growing by 2 years age.

Neurological-hydrocephalus, convulsion, respiratory-pulmonary hypertension, cor pulmonale, cardiac-cardiomyopathy, endocardial fibroelastosis, valvular regurgitation, heart failure. Gastrointestinal-macroglossia, umbilical and inguinal hernia (early symptoms), enlarged liver and spleen due to GAG deposition, functioning is normal. Corneal clouding, hearing loss, hair is coarse and abundant.[6,8,9]

Developmental delay seen by age 1-2 years, maximum functional age of 2-4 years. There is progressive deterioration.

In the present case there was no family history of Hurler syndrome.

Diagnosis is made by-urinary GAG levels, enzyme activity assays, gene sequencing and prenatal diagnosis.

Treatment

Enzyme replacement therapy with recombinant human alpha L-iduronidase (Aldurazyme) given weekly intravenous injection, hematopoietic stem cell transplant ideal for patients under 2 years of age.[10] Supportive and surgical interventions, gene therapy-under research.

CONCLUSION

Hurler syndrome is an autosomal recessive disorder due to deficiency of enzyme alpha L-iduronidase (IUDA) causing accumulation of GAG in tissue. Average age of

mortality is 5 years. The present case of Hurler syndrome is an unusual diagnosis which should be kept as a differential diagnosis in cases of delayed milestones and coarse facial features, clouding of cornea, umbilical hernia, skeletal abnormalities.

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