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A Case Report On Hurler Syndrome

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Abstract

Mucopolysaccharidoses are hereditary, progressive diseases caused by mutations of genes coding for lysosomal enzymes needed to degrade glycosaminoglycans (acid mucopolysaccharides). Failure of this degradation because of absent or grossly reduced activity of mutated lysosomal enzymes results in the intralysosomal accumulation of glycosaminoglycan fragments. (1)

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. (2)

Incidence of MPS I-H has been reported to be 1:100,000 per child birth, and no predilection for sex and ethnicity has been found. (3)

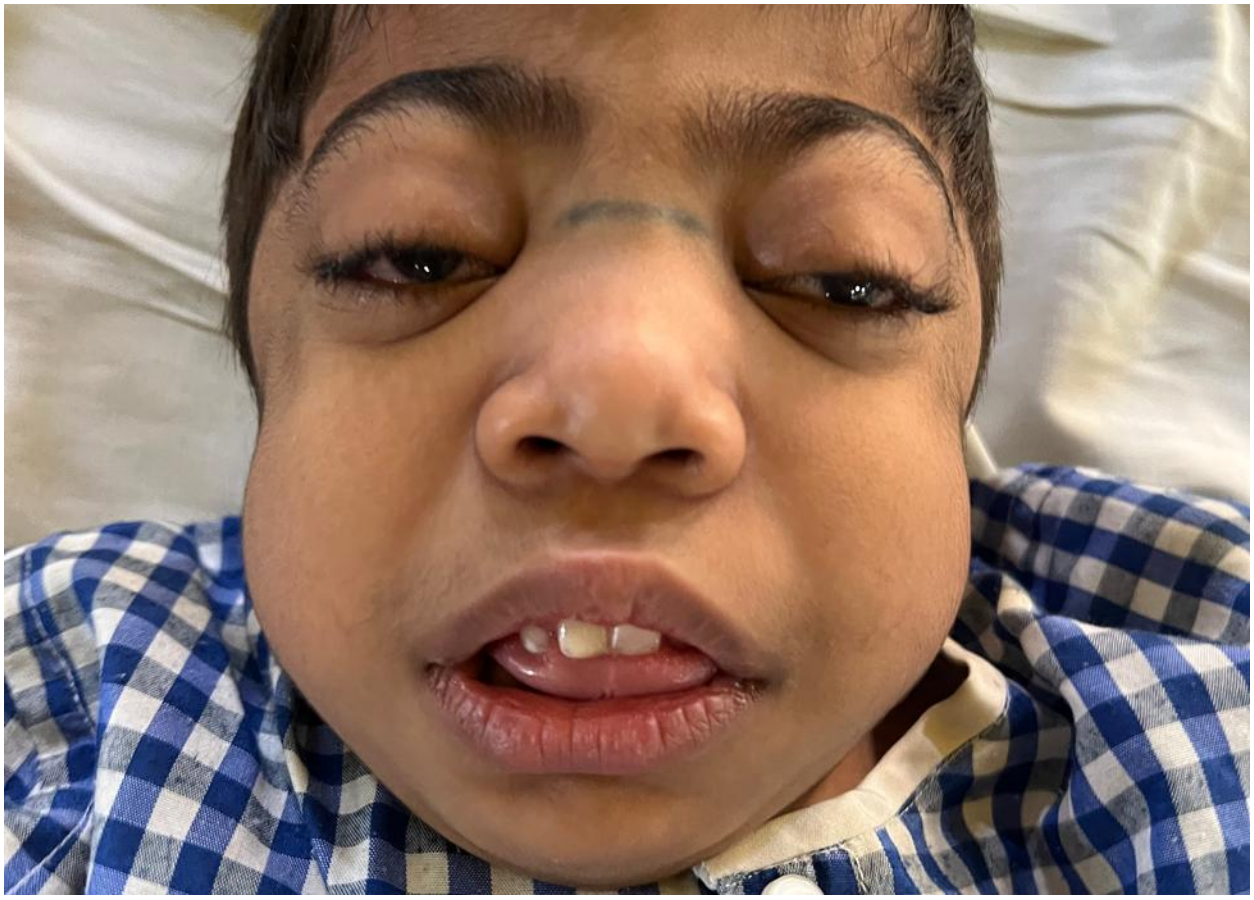
Hurler syndrome (MPS I - H) is the most common and severe form of mucopolysaccharidosises. Deficiency of this enzyme results into a wide range of phenotypes including Hurler's (severe), Scheie's (mild) and Hurler-Scheie (intermediate) syndromes. (4)

Keywords: Mucopolysaccharidoses, Glycosaminoglycans, Alpha-L-iduronidase enzyme, Hurler syndrome, Umbilical hernia, Inguinal hernia, Delayed milestones, Nystagmus, Cloudy cornea, Optic disc pallor.

Introduction

Case report-

A 5year old female child, born of consanguinous marriage (third order) presented to paediatric opd with complaints of swelling over umbilical and inguinal area present since one year which gradually increasing in size but non tender and reducible.



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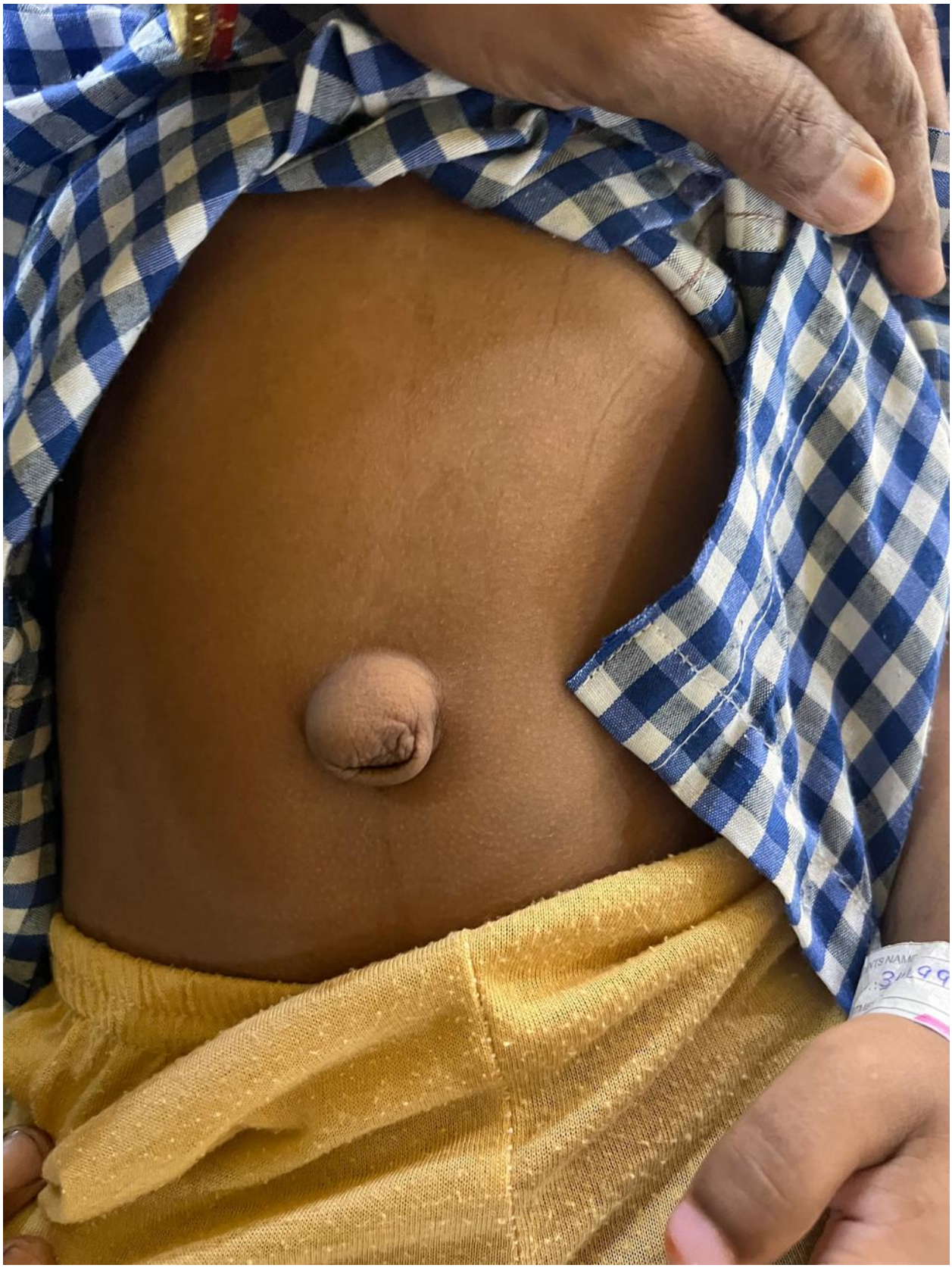

On examination- Patient had delayed milestones, inability to walk,coarse facial features, flattened face,slurred speech, webbed neck, recurrent history of Upper respiratory tract infection (7-8 times a year), grade two clubbing, stunting and wasting present, dolicocephaly, low hair line, low set ears, bilateral moderate to severe hearing loss, mouth breathing present, broad and short nose with depressed nasal bridge, large fissured tongue,

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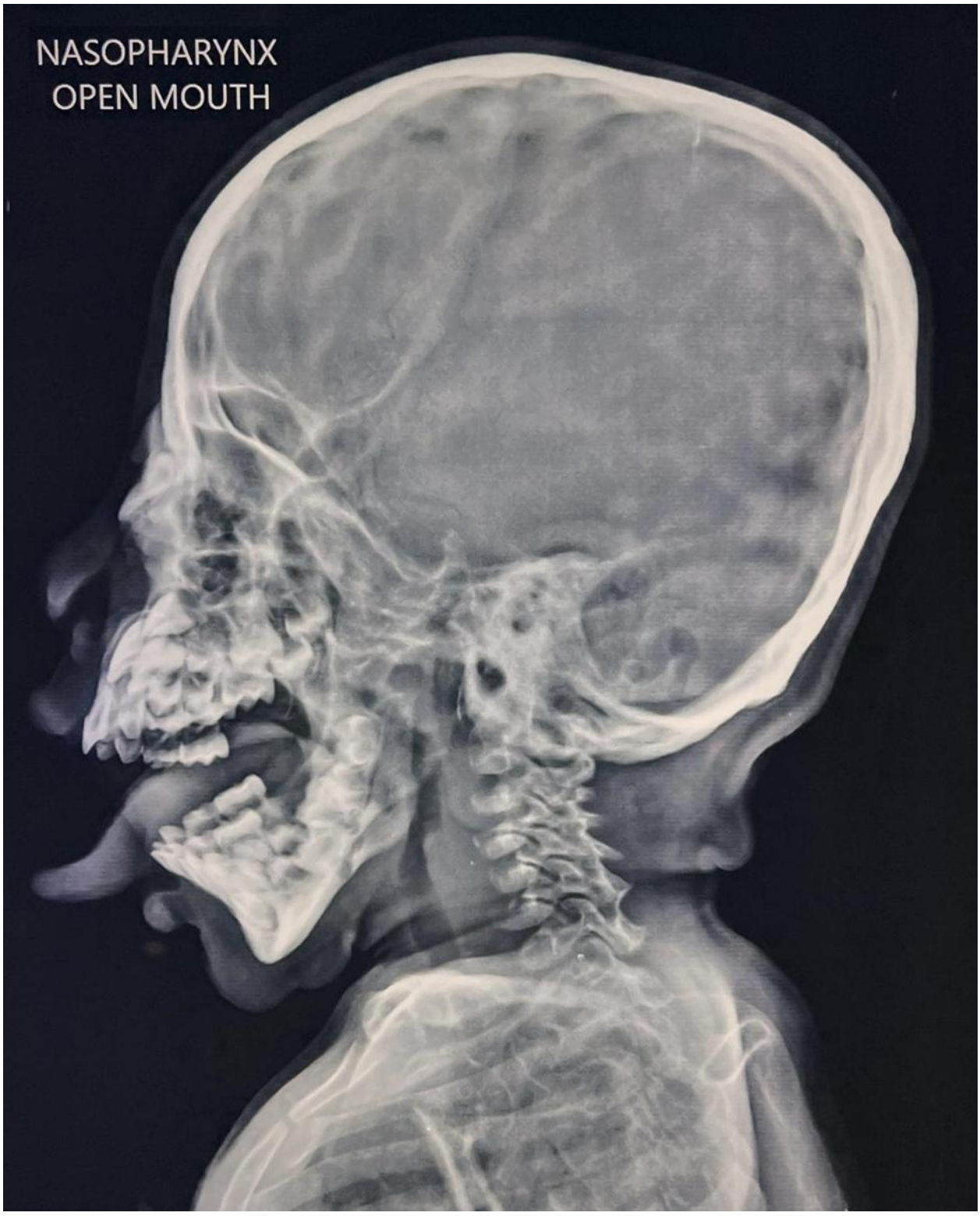
bilateral grade one tonsillar enlargement, Micrognathia, asymmetry of chest present with left precordial bulge, bilateral cubitus valgus and bilateral genu valgus and sacral dimpling present. On CNS examination- tone and muscle power was normal.

Full body X-ray was advised which came with multiple positive findings including-

Skull- macrocephaly with "silver beaten appearance" , J shaped sella Tursica, facial bones appears hypoplastic with crowding of the teeth, non pneumatisation of the mastoid is seen.



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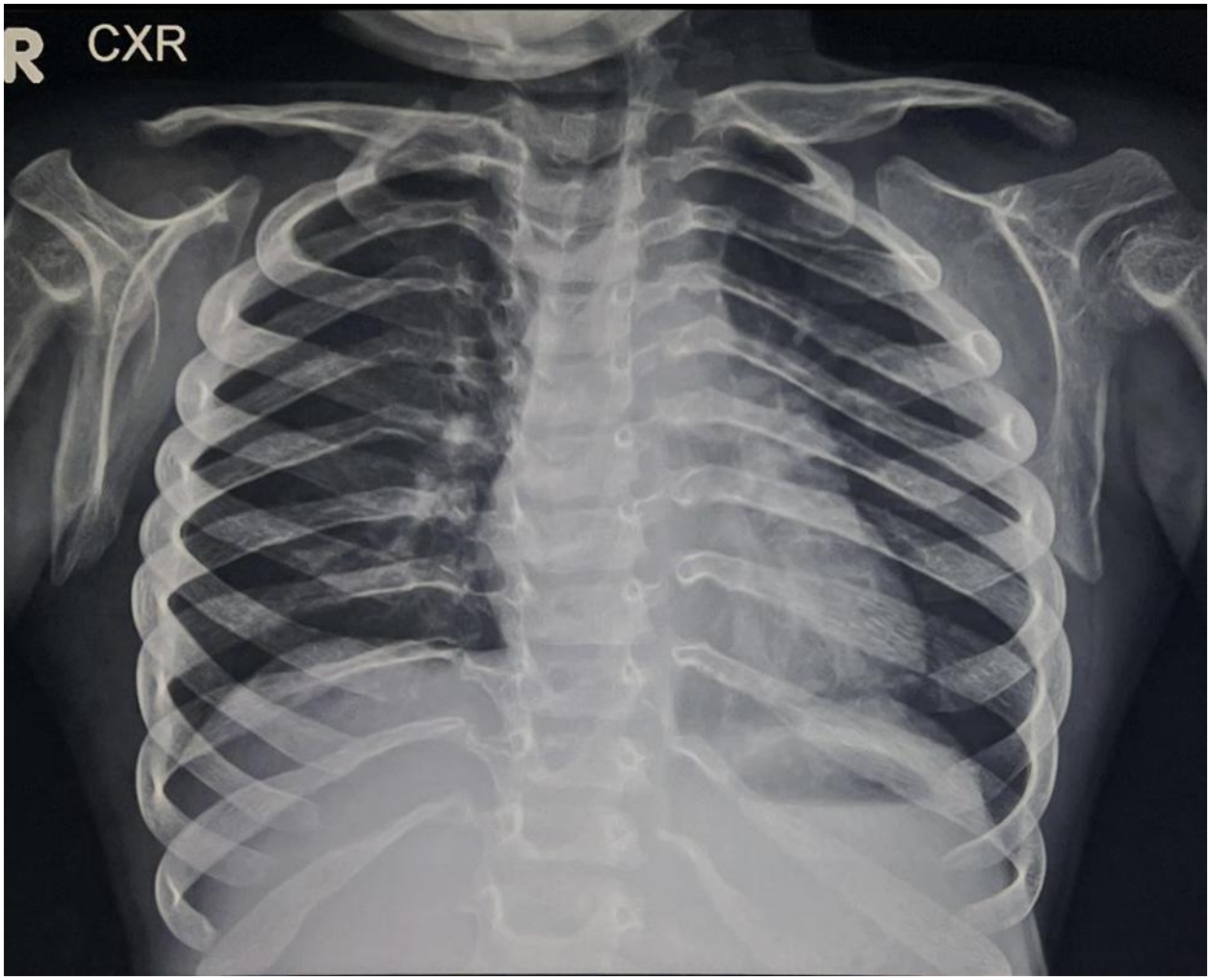

Neck- Odontoid appears hypoplastic with Atlanto axial subluxation is seen

Chest- both clavicles and the bilateral lower ribs (10th and 11th) appear "OAR SHAPED", ribs were widely spaced.

Spine- mild platyspondyly in thoraco lumbar region with "antero inferior beaking" and posterior scalloping, anteriorly bowed sternum (Paetus Carinatum)

PBH- Flaring of the iliac wings with narrow iliac base and shallow acetabulum.

Long bones- Upper limb- Humerus, Radius and ulna appears short and wide.

Distal ends of radius and ulna appears dysplastic with a V-shaped configuration. Metacarpals show proximal pointing (except for thumb)

Proximal phalanges appear "bullet shaped".

Lower limbs- long bones show no obvious abnormality.

There is evidence of retarded skeletal maturity is seen as the bone age is 2-3 years.

The "antero-inferior beaking of the vertebral bodies" also confirms the diagnosis.

On USG ABDOMEN- Small completely reducible hernia present.

On BERA- Moderate to severe hearing loss present.

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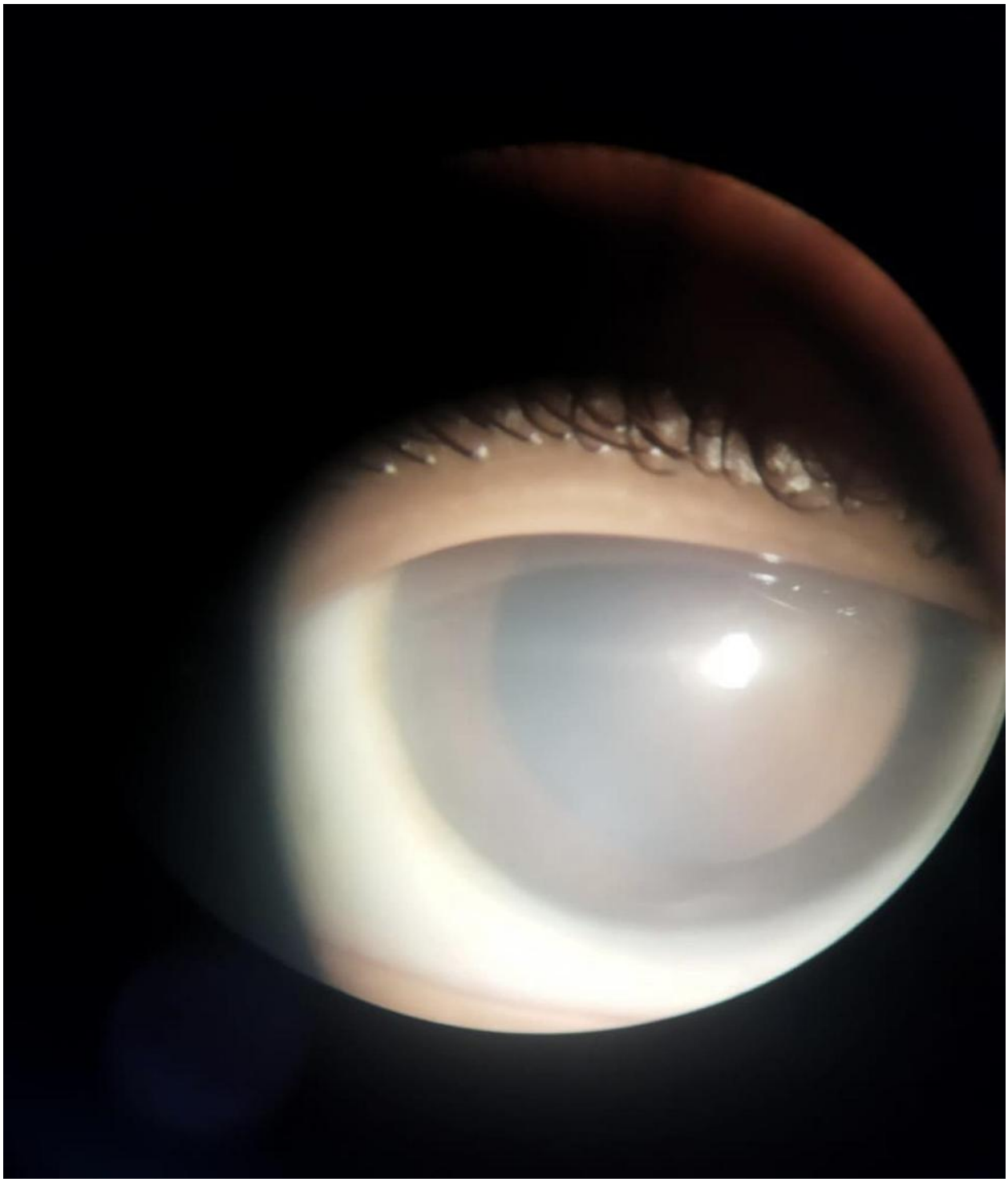
On 2D echo- Myxomatus mitral valve, Mild Mitral regurgitation, mild thickening of aortic valve with hypertrophy of left ventricle.

Patient was referred to Ophthalmology department for ocular assessment. On examination-

Vision of the patient was central, unsteady, unmaintained with right face turn. On retinoscopy, high sphere of -4 with cylinder of -1 was prescribed to the patient for both eyes. On slit lamp examination pupils were sluggishly reacting to light, nystagmus was present which was horizontal pendular with small amplitude and medium frequency, shallow orbits and cloudy cornea (grade3) was present. On dilated fundus examination- both eye optic disc pallor was present (?optic atrophy) , posterior pole vessels were tortuous and foveal reflex was dull.



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All these features are suggestive of the HURLER SYNDROME.

To confirm the diagnosis genetic study was advised which came with the positive report.

Discussion- Hurler's syndrome is caused by deficiency of the lysosomal enzyme, α-L-iduronidase, is traced to the chromosome 4p16.3 (5)

The clinical manifestions include short stature, mental retardation, enlarged skull, low nasal bridge, thick coarse facies, deafness, short hands and feet with vertebral anomalies like atlantoaxial subluxations [6], inferior beaking of vertebral bodies and infiltration of duramater and cervical cord with Mucopolysaccharides [7]. Storage disorders that produce skeletal abnormalities are collectively termed as "dysostosis multiplex." Cardiovascular manifestations like cardiomyopathy, pulmonary arterial hypertension and heart failure are commonly noted in MPS1. Frequent upper and lower respiratory tract infections are commonly encountered secondary to enlarged tonsils, adenoids. Most often the affected children usually die within 1st decade due to either cardiac or respiratory failure. The diagnosis of MPS1 is made by finding out lysosomal enzyme, α-L-iduronidase, levels in plasma, peripheral leucocytes and cultured fibroblasts. In our case, most of the findings classically described in literature are found out. Enzyme replacement therapy consists on intravenous administration of the recombinant alpha-L- iduronidase enzyme at regular intervals, which reduces lysosomal overload and thus improves some clinical signs[8]. But due to high cost and low neurological efficacy (does not cross the haematopoietic barrier), HAEMATOPOIETIC STEM CELL TRANSPLANTATION is a treatment of choice especially when performed early before the onset of psychomotor developmental delay. (9)

These treatments should be combined with psychological support and genetic counseling. The genes of the different enzymes involved are now located and many mutations have been identified. Prenatal diagnosis is possible on culture of amniotic cells.(10)

When there is positive family history of Mucopolysaccharidosis, genetic counseling and testing for the enzyme should be recommended for the newlywed families.

Conclusion- This case report, we have come to the diagnosis of Hurler's Syndrome, Mucopolysaccharidosis type 1 by the clinical manifestations and radiological evidence of specific features.

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