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Hurlers –Scheie's syndrome with Dextrocardia: A Rare Presentation

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Abstract:*A 24 year female with clouding of cornea, coarse facies, gapping of teeth, large tongue, claw hand, hepatomegaly, umbilical hernia was diagnosed as a case of Hurler-Scheie Disease. An unusual feature was that the patient was having Dextrocardia, not reported yet as cardiac feature of Hurler-Scheie's Disease. An Ophthalmologist should examine not only eyes but also systematically to come to a final diagnosis in such rare disease with rare feature.*

Key Words:*clouding of cornea, Hurler's disease, Scheie's disease, mucopolysacchridosis, Dextrocardia*

I. Introduction

Mucopolysaccharidosis (MPSs) are inheritable storage diseases caused by a deficiency of lysosomal enzymes that degrade glycosaminoglycans (GAGs, previously called mucopolysaccharides). The MPSs are a heterogeneous group characterized by the intralysosomal accumulation of GAGs, excessive urinary excretion of GAGs, and variable degrees of progressive mental and physical deterioration and, in severe forms, premature death [1]Hurler-Scheie's syndrome is an autosomal recessive mucopolysaccharidosis which results in reduced activity of the enzyme a-L-iduronidase with subsequent accumulation in the tissues of heparan and dermatan sulphate. The syndrome represents a spectrum of disease, patients with the more severe, rapidly progressive disease being classified as Hurler's syndrome (mucopolysaccharidosis I-H) and those less severely affected as Scheie's syndrome (mucopolysaccharidosis I-S). [2]

Hurler-Scheie describes a clinical phenotype that is intermediate between Hurler and Scheie syndromes and is characterized by progressive somatic involvement [1]

Incidence of Hurler-Scheie disease is 1:500,000 [3,4]

Clinical features of Hurler's –Scheie's syndrome includes: [5,6]

Non-ocular features

1. Abnormal facies, enlarged skull, thickened lips, prominent forehead, coarse hair, abnormally low ears
2 . Macroglossia, Hirsutism
3. Contracture of hips, knees, elbows, and fingers
4. Hepatosplenomegaly
5. Diastasis recti, umbilical hernia
6. Growth retardation, mentally deficient, slow psychomotor growth
7. Mental retardation
8. Heart defects

Ocular features

1. Clouding of the cornea: the opacities are stromal and Bowman's membrane is absent
2. Defective dark adaptation and ERG
3. Retinal degeneration
4. Optic atrophy
5. Ptosis
6. Strabismus (Esotropia)
7. Glaucoma

The cardiac involvement in individuals with MPS I HS (Hurler-Scheie) & MPS I S (Scheie) Syndromes are usually milder and survival into adulthood is common.

The cardiac valves (most often the mitral and aortic valves) are thickened by the deposition of glycosaminoglycans and these valves can either leak (valve regurgitation) or becomeobstructive to flow (valve stenosis).

The heart muscle itself can become thickened and less compliant than normal because of glycosaminoglycan deposition.

Most importantly, the coronary arteries – the vessels that supply blood to the heart muscle itself – can be diffusely narrowed by glycosaminoglycan deposition[7]
Infiltration of the heart's conduction system by glycosaminoglycan can cause the development of a potentially lethal heart rhythm disturbance (complete heart block) and require placement of permanent pacemaker.

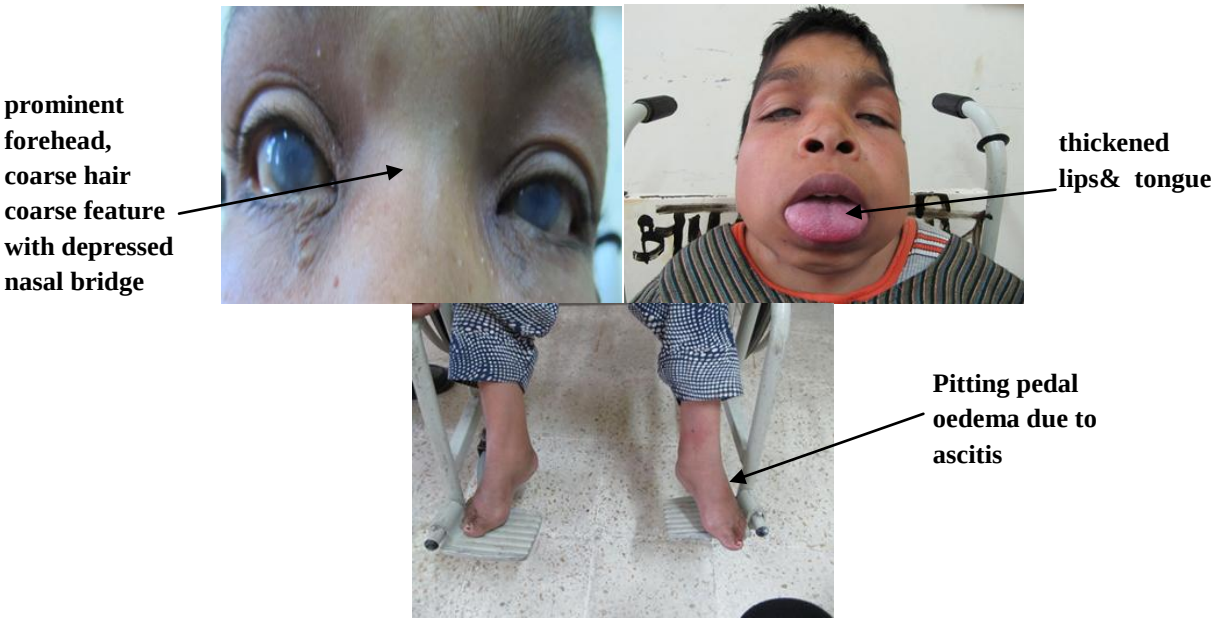
The various cardiac anomalies in a patient of Hurler's- Scheie's syndrome includes mitral regurgitation, Aortic valve disease, conduction defects, heart block, congestive heart failure.[1,6,7]
Thus the above mentioned cardiac manifestations in patient with Hurler's-Scheie's syndrome warrants through cardiac work up and follow up.

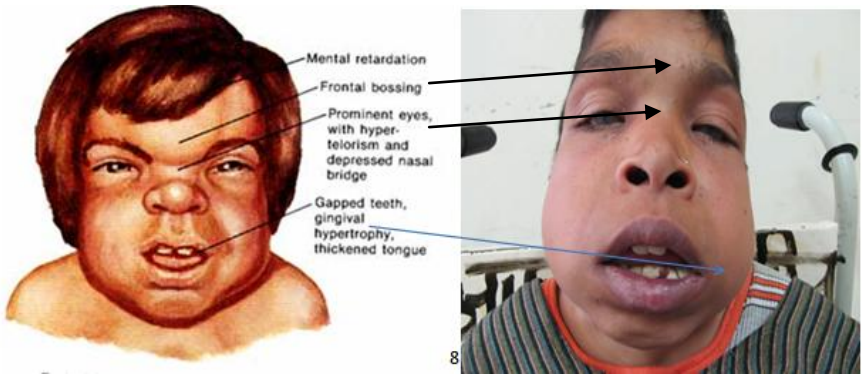
We reported a rare case of Hurler's- Scheie's syndrome with dextrocardia, probably not presented till now/ reported with other associated features.
Case report:

A 24 year female resident of uttarakhand, india was referred to department of ophthalmology for diminution of vision BE *1 yr.
The child was born of a nonconsanguineous marriage and was the first child of the couple.According to the parents, all her milestones were delayed. On examination she was having puffiness around the lids, cornea was cloudy sparing about 1mm in the centre that too was hazy. AC was deep, pupil was regular, round and reacting, phakicstaus of lens was present. Rest details were not clear due to corneal haze. Snellen's visual acquity was OD:1/60, OS:1/60.IOP AT OD :22mm Hg, OS: 22 mm HgInitially we made congenital glaucoma, congenital hereditary endothelial dystrophy,and mucopolysacharidosis as provisional diagnosis.On further local and systemicevaluation we found following findings.Patient corneal diameter was within normal range, no such feature were present in other family members.

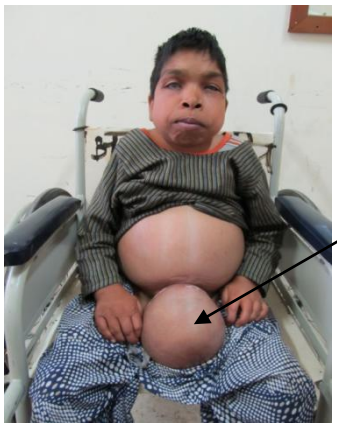
On systemic evaluation we find:
1.Patient was well oriented to time , place and person, having short stature(3ft 5 inch), and low IQ.
2. Enlarged skull, thickened lips, prominent forehead, coarse hair coarse feature with depressed nasal bridge
3. Gapped teeth, gingival hypertrophy, thickened tongue was present.
4. Patient was having features of joint stiffness skeletal deformity, dysostosis multiplex.
5. Claw hand, and large umbilical hernia was present
6. Patient was having ascites, shiny abdominal skin with prominent superficial veins.

Patient menstrual history…
BP= 130/68mmHg, PR=76 ,
SI, S2were normal heard on right side of the heart.
On abdominal examination hepatomegaly was appreciated.





Claw hand in Hurler-Scheie's syndrome



Umblical Hernia in Hurler-Scheie's syndrome

On investigation:

Haemogram:

Hb=14.3 gm/dl, TLC= 6100/mm3,DLC=N79L20M01, RBS=54gm/dl, urea =31gm/dl, creatinine=0.6 gm/dl, SGPT=14, SGOT=24, ALP=249, Bilirubin total=1.4, direct bilirubin=0.6, indirect bilirubin= 0.8

Total protein=6.8, albumin =3.8, globulin=3.0, ESR=04mm in 1st hr, GBP= normochromic with mild anisocytosis.

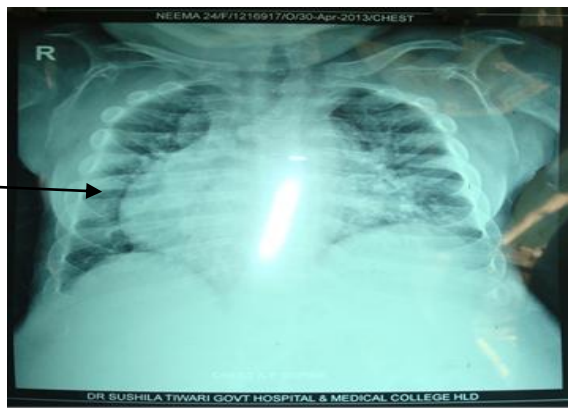
24 hr urine protein= 60mg(10-15mg per 24 Hr)

<u>On peritoneal tap:</u>

Amount=3.0 ml, colour = yellow, PH=6.0, protein =3.0, sugar/glucose=122, cells =25, differential count P30L70(background has foamy and reactive mesothelial cells)

XRay , and ECHO confirms the Dextrocardia.

Dextrocardia (situs inversus)



USG whole abdomen suggestive of:

Hepatomegaly without focal lesion (non specific)

Non visualization of uterus along with adnexal region due to large herniated sac in the pubic symphysis.

Spleen , kidney, pancreas, gall bladder were within normal limits.

History as given by patient and patient relatives, clinical evaluation and investigations lead us to make Hurler-Scheie's Disease with Dextrocardia as diagnosis.

II. Discussion

Hurler-Scheie is a autosomal recessive condition. The onset of symptoms is usually observed between 3–8 yr; survival to adulthood is common. Cardiac involvement and upper airway obstruction contribute to clinical mortality. Patients have micrognathia and spondylolisthesis, which may cause cord compression. [1]

This patient is having most of the clinical features with one rare feature of dextrocardia. In standard texts it is not written as a cardiac manifestation of Hurler's-Scheie.

This is an important feature as the most commonly death occurs in Hurler –Scheie's is due to cardiac-respiratory diseases.

So, it is customary to evaluate the cardiac status of the patient and keep in view this rare cardiac manifestation which can be present in such patient's.

Treatment includes bone marrow transplantation which results in significant clinical improvement of somatic disease in MPS I and increased long-term survival. Resolution or improvements have been noted in hepatosplenomegaly, joint stiffness, facial appearance, obstructive sleep apnea, heart disease, communicating hydrocephalus, and hearing loss. However, the skeletal and ocular anomalies are not corrected

Enzyme replacement using recombinant enzyme is a promising method to treat the somatic features of MPSs. There are ongoing clinical trials of recombinant a-L-iduronidase for MPS I patients.

Supportive management, with particular attention to respiratory and cardiovascular complications, hearing loss, carpal tunnel syndrome, spinal cord compression, and hydrocephalus, can greatly improve the quality of life for patients and their families.[1]

From ophthalmological point of view we referred the patient for penetrating keratoplasty at higher centre, to be followed up in our department.

Sometimes an ophthalmologist may become a news breaker for a life threatening disease. And as it is well said '**Eye is a mirror of the whole body**' so, it is essential for an ophthalmologist to evaluate the entire body from head to toe, as we have done in this case to reach a final diagnosis of an unusual case.

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