HURLER SYNDROME - A CASE REPORT

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INTRODUCTION

A 5 year old male child reported to the department of Pediatrics with large head, facial puffiness, enlarged abdomen and corneal opacity.

Examination revealed that the child had stunted growth and a short neck. Shape of the head was dolichocephalic with marked macrocephaly. Coarse facial features like depressed and broad nasal bridge, flaring of both nostrils, prominent supra orbital rim bilaterally, facial asymmetry was more marked on the right side due to diffuse swelling at angle and body of mandible. Child had an enlarged abdomen, herniated umbilicus and massive hepatosplenomegaly. Both the hands were short and stubby. There was history of delayed milestones and appeared to be disabled intellectually.

IMAGING WORK-UP

X rays of the skull, limbs and spine was done which suggested the diagnoses.

FIG 1 X RAY SKULL LATERAL VIEW

shows enlarged size of cranium and J shaped sella

X RAYS B/L HANDS AP AND LS SPINE LATERAL VIEW

Fig 2(A) Shows pointing of the proximal ends of the metaphyses

Fig 2(B) Shows inferior beaking of the vertebrae
Fig 3 (A) & (B) show widening of the anterior ends of the ribs.

The cardiac shadow appears normal.

DIAGNOSIS

- On the basis of above mentioned clinical features and presented x-rays, a provisional diagnosis of mucopolysaccharidosis (Hurler’s syndrome) was made.
- After enzymatic analysis, the diagnosis was confirmed.
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DISCUSSION

- The MPS disorders are inherited as a recessive trait and are manifested by progressive changes evident in late infancy or early childhood.
- Each type of MPS is identified by a specific enzymatic defect; however, the skeletal findings are similar in several of these abnormalities.
- Hurler’s syndrome is one form of mucopolysaccharidosis (MPS type II), a group of several storage diseases that produce skeletal abnormalities collectively termed “dysostosis multiplex”.
- It is named after Gertrud Hurler, a German paediatrician (1889-1965)
- The estimated incidence is at ~1:100,000.
- It carries an autosomal recessive inheritance and manifests in the first years of life.
- The specific enzymatic defect in Hurler’s syndrome is alpha 1-iduronidase.

MANAGEMENT

Specific treatment or cure is limited for MPS. Management has been limited to supportive care and experimental treatment modalities. Medical treatment modalities include the following:

- Laronidase
- Idursulfase
- Elosulfase alfa

Surgical care for specific conditions may include the following:

- Hydrocephalus – Ventriculoperitoneal shunting
- Corneal clouding – Corneal transplantation
- Cardiovascular disease – Valve replacement
- Obstructive airway disease – Tracheostomy
- Orthopedic conditions – Carpal tunnel release; soft tissue procedures to release hip, knee, and ankle contractures; hip containment surgeries; corrective osteotomy for progressive valgus deformity at the knee; posterior spinal fusion
REFERENCES


