Hurler’s Syndrome

K. CHATTERJEE, R. R. PAL, M. MAJUMDAR, A. MANKA & A. M. SAHA

Hurler’s Syndrome is the prototype of a group of genetic disorders, involving disturbances in mucopolysaccharide metabolism. A case of Hurler’s Syndrome having rare association with otologic and stomatologic disorders, is presented herewith, along with the relevant clinical, radiological and biochemical findings.

The mucopolysaccharidoses are lysosomal storage diseases. They result from deficiency of specific lysosomal enzymes involved in the degradation of dermatan sulfate, heparan sulfate, or keratan sulfate singly or in combination. Incompletely degraded mucopolysaccharides accumulate in tissues and are excreted in the urine. Presently, ten enzymes are known, deficiency of any one of which can result in a specific mucopolysaccharide storage disease. Diagnosis is based on the clinical and radiological findings, the type of inheritance and especially on the particular mucopolysaccharides to be found excreted in the urine and accumulated in the body cell.

The prototype is the Hurler’s Syndrome or mucopolysaccharidosis I (MPS I), which is inherited as an autosomal recessive trait. Hunter’s Syndrome (MPS II) is a less severe condition which is X-linked recessive.

The rarity of Hurler’s Syndrome having its unusual association with otologic and stomatologic abnormalities prompted us for publication of this case.

Case Report

A 10-year old boy (H. No.—9861) presented to the out patient department of R. Ahmed Dental College & Hospital, Calcutta, with complaint of severe pain and swelling in the left lower jaw. He also had the complaints of inability to close the fist since childhood and the recent history of impairment of hearing with ear discharge, night blindness and shortness of breath on exertion.

General examination

Short stature, large head with a prominent forehead, coarse features, hypertelorism, flat nasal bridge, large nostrils, bilateral diffuse corneal clouding (Fig. 1), short arms, small fingers and palms with typical ‘Claw hand’ (Fig. 2), firm skin with hypertrichosis, pot belliied abdomen with hepatosplenomegaly and umbilical hernia (Fig. 3), delayed mile stones with impaired I.Q.

Case Report

A 10-year old boy (H. No.—9861) presented to the out patient department of R. Ahmed Dental College & Hospital, Calcutta, with complaint of severe pain and swelling in the left lower jaw. He also had the complaints of inability to close the fist since childhood and the recent history of impairment of hearing with ear discharge, night blindness and shortness of breath on exertion.

![Fig. 1: Showing typical facies with Corneal Clouding.](image1)

The rarity of Hurler’s Syndrome having its unusual association with otologic and stomatologic abnormalities prompted us for publication of this case.

Intra-oral examination

Presence of following teeth

<table>
<thead>
<tr>
<th>6 EDC 21</th>
<th>12 CDE 6</th>
</tr>
</thead>
<tbody>
<tr>
<td>6 EDC 21</td>
<td>12 CDE 6</td>
</tr>
</tbody>
</table>

![Fig. 2: Showing “Claw Hand”.](image2)

![Fig. 3: Showing Pot-bellied abdomen with umbilical hernia.](image3)

Teeth present were of normal size and shape without any malocclusion and with slight hypertrophy of gingiva due to poor oral hygiene. The patient was clinically diagnosed as a case of Mucopolysaccharidoses. For his present dental problems he was treated with antibiotic syrup followed by extraction of E E along with an incisional biopsy of gingiva from the region of E. He was subsequently referred to E.N.T.O.P.D. of Medical College Hospital, Calcutta, for E.N.T. problems and was further advised for radiological and cardiological check up, routine and specific biochemical investigations of blood and urine.

E.N.T. examination

Bilateral central perforation with discharge—moderate in amount, mucoid in nature and non-offensive. Rinne Test showed early conductive type of deafness more in the right side. Tonsils were enlarged, good

![Reprint requests to:](image4)
sized adenoids present in the nasopharynx causing airway obstruction.

Radiological examination

Chest—P.A. view, Skull including Mandible—A.P. and Lateral, Dorsal spine—A.P. and Lateral, Pelvis including Lumbar spine—A.P. and Lateral. Both hands—A.P. view, Law's Lateral view and Town's view were done. Careful evaluation of radiographs revealed—dolichocephalic skull, slightly elongated sella turcica, normal jaw bones without any dilatation of dental follicle (Fig. 4), hump like deformity in the dorsal and upper lumbar vertebral bodies along with anterior beaking (Fig. 5), small, poorly developed carpal epiphyses, proximal tapering of the metacarpals and distal tapering of the proximal phalanges (Fig. 6) and sclerotic type of mastoids.

Cardiological Examination

E.C.G. and Echo-Cardiogram were within normal limits and radiographically there was no cardiac abnormality.

Histopathological examination of gingival biopsy

Sections stained with haematoxylin & eosin—

Slightly increased thickness of the prickle cell layer of superficial epithelium and moderately increased collagenisation of the underlying lamina propria (Fig. 7).

Fig. 4: Showing radiograph of jaw and tooth follicle.

Fig. 5: Showing hump like deformity of the vertebral body.

The overall radiological features were suggestive of Mucopolysaccharidosis I (MPSI).

Haematological Examination

Almost all the reports of the haematological investigations T.C., D.C., E.S.R., Hb%, Sugar P.P., Cholesterol, Triglycerides, Total serum protein, Electrophoretic pattern of serum protein were within normal limits.

The study of the lysosomal enzyme from the patient's and normal leucocyte showed the following:

\[ \text{iduronidase (} \mu \text{ mol/gm.} / \text{protein hr.)} \]

<table>
<thead>
<tr>
<th>Patient</th>
<th>Normal</th>
</tr>
</thead>
<tbody>
<tr>
<td>(1)</td>
<td>30</td>
</tr>
<tr>
<td>(2)</td>
<td>48</td>
</tr>
</tbody>
</table>

(Normal range 15—34)

Urine Examination

Special urinary investigations were done for the diagnosis of mucopolysaccharidoses.

Total Excretion of M.P.S.—

42 mg uronic acid/day.
(Normal range at this age—2-5 mg/day)

The excreted glycosaminoglycans which was purified after dialysis was either dermatan Sulfate or heparan sulfate. It was confirmed by hyaluronidase treatment. At this was not hydrolysed by hyaluronidase, it was either dermatan sulfate or heparan sulfate.

It was further confirmed by acid hydrolysis and paper chromatography of purified glycosaminoglycans from urine showed that GAG contained mostly galactosamine indicating the presence of high concentration of Dermatan sulfate in comparison to heparan sulfate which was the characteristic finding of Hurler's Syndrome. Dermatan sulfate : Heparan sulfate = 7:3.

The clinical diagnosis was thus supported by biochemical analysis and the present case was established as mucopolysaccharidosis I (MPSI) or Hurler's Syndrome.