Maroteaux -Lamy Syndrome: Report of Two Cases in Siblings

Mucopolysaccharidosis (MPS) is a rare autosomal recessive inherited glycosaminoglycan storage disease caused by the deficiency of arylsulfatase., also known as N acetyl glucosaminoglycans[1]. It takes its name from the two French doctors Maroteaux and Lamy who first described the condition in 1963[2]. It has been estimated that about 1 in 1,300,000 births are affected by MPS. Deficiency of Aryl sulfatase-B leads to accumulation of dermatan sulfate in tissues and their excretion in urine[3]. At least 3 distinct ages of onset have been differentiated[4]

- A severe infantile form, characterized by early onset and rapid disease progression
- An intermediate type is characterized by onset of disease in late childhood and
- A mild or adult form demonstrates onset after second decade.

Disease progression in the juvenile and adult form istypically slower than in infantile form.

Affected individual usually have retarded growth ranging from 90 to 140 cm, intelligence is not affected, large head, short neck, chubby cheeks, broad nose with flat bridge and wide nostrils. The shoulders are narrow and rounded and the stomach tends to protrude[2,3]. The hair on the body is coarser and more abundant than usual, and the eyebrows are bushy. Skin may become thickened and less elastic than usual.[5] Neck is short contributing to breathing problems. Individuals with MPS may end up with secondary bacterial infections. Thick lips and enlarged tongue, broad alveolar ridges, widely spaced teeth with fragile enamel are some of intraoral features. Individuals with the syndrome may develop heart failure and may have problem withaortic and mitral valve.[6]

General manifestations include hepatosplenomegaly, umbilical or inguinal hernia, bowel problems like diarrhea significant problems with bone formation and growth called dysostosis multiplex, spine abnormalities like kyphosis, scoliosis, joint stiffness, short and broad hands with stubby fingers.[4,5,6] Fingers stiffen and gradually become curved due to limited joint movement giving claw like appearance. Many people with MPS VI stand and walk with their knees and hips flexed.[7] These combined with a tight Achilles tendon may cause them to walk on their toes and sometime have knock knees.[7,8] Cloudy corneas due to storage of GAG, conductive deafness and carpel tunnel syndrome due to compression of nerve.[9] (Fig. 1)

Case Report:

A 11-year male patient and female patient as a twins of normal intelligence of aconsanguineous married parents came to the Outpatient Department with the chief complaint of his retained root stumps. Both were suffering from respiratory obstructive disorders and cardiac valvular disease (mitral valve) since early child hood. The girlpatient had blurred vision since 4 years. General physical examination revealed noisy breathing andhigh respiratory rate of 24 cycles/min in female patient and 27 cycles/min in male. Patients had distended abdomen with umbilical hernia, retarded growth (height being 142 cm of female patient and 140 cm of male patient) restricted joint movements of elbows and phalanges. Patient had spine abnormalities with droopy shoulders. (Fig.2) Patients had distended abdomen with umbilical hernia, retarded growth (height being 142 cm of female patient and 140 cm of male patient) restricted joint movements of elbows and phalanges. Patient had spine abnormalitieswith droopy shoulders. Extraorally patients had large head, macrognathia, frontal bossing, and saddle nose with wide nostrils, coarse and bushy eyebrows, coarse facial hair, cloudy corneas and short neck. (Fig.3)

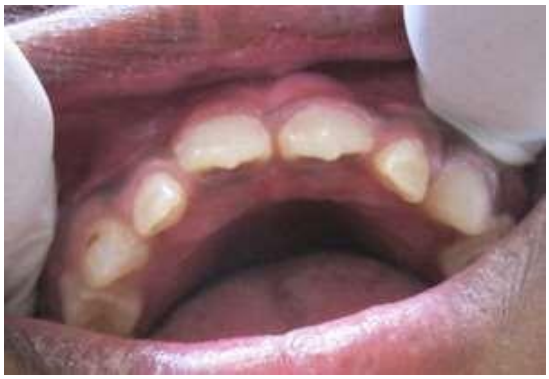

Figure 1 showing stunted growth, Bow like legs, Claw like fingers

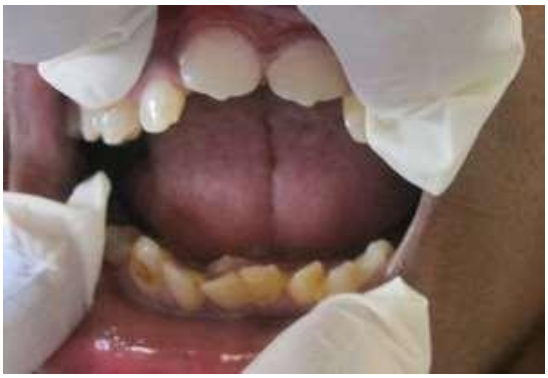

Figure 2 showing Flarred nostril, chubby cheek

Patients had distended abdomen with umbilical hernia, retarded growth (height being 142 cm of female patient and 140 cm of male patient) restricted joint movements of elbows and phalanges. Patient had spine abnormalitieswith droopy shoulders. Extraorally patients had large head, macrognathia, frontal bossing, and saddle nose with wide nostrils, coarse and bushy eyebrows, coarse facial hair, cloudy corneas and short neck. (Fig.3)

Figure 3 showing depressed nose, Frontal Bosing, Widened eyebrows

Intraorally patients had high arched palate with linear grooving at the center, macroglossia, fissured tongue, spacing between teeth, open bite, with over retention of deciduous dentition in the female patient.

Figure 4 showing intra oral photos

Unerupted anterior teeth, incomplete root development of permanent anterior teeth, condylaraplasia, prominent gonial angle and increased follicular spaces having dentigerous cyst like appearances around unerupted molars.(Fig.5)

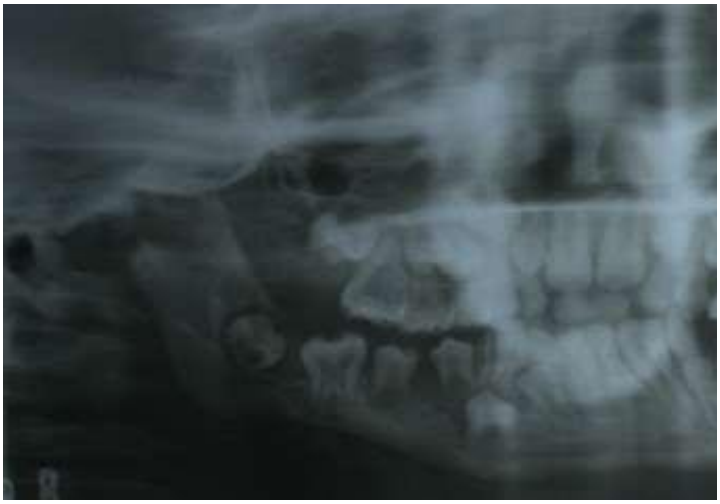

Figure 5 Demonstrating Panoramic Radiograph

Systemic examination revealed hepatosplenomegaly. Patients were referred by pediatrician for lab investigations for urine analysis which revealed abnormally high levels of GAG concentration in urine which was 594.70 mg gag / g creatinine (normal= 19.97- 110.53). Enzyme assay revealed abnormally low levels of aryl sulfatase B that was 15.42 N mol/hr(normal >120 N mol/hr)which is pathognomonic sign of MPS.

- **Treatment plan:**

1. Prompt treatment of respiratory infections from pulmonologist

2. Prophylaxis against infective endocarditis before any dental or surgical procedure.

3. Annual checkup with the cardiologist.

- **Dental Care:**

1.Extraction of root stumps in relation to 84

2.Preventive care: flouride application

3.Maintenance of oral hygiene

4.Regular dental check up

Conclusion:

Although Maroteaux-Lamy syndrome is considered rare, these disorders are devastating for individuals and their families and result in considerable use of resources from healthcare systems; however the magnitude of the problem is not well defined.Theintroduction of Enzyme replacement therapy with Galsulfase has been a milestone in the treatment of MPS patients. This therapy opens the door to a more proactive approach of managing the disease, i.e., slowing down the accumulation of GAG rather than alleviating the resulting clinical manifestations.

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