

MORQUIO DISEASE : A CASE REPORT

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Summary

Mucopolysaccharidoses (MPS) are hereditary progressive metabolic disorder caused by the absence or malfunctioning of lysosomal enzyme needed to breakdown molecule called glycosaminoglycan (GAGS) [1]. The major GAGS are chondroitin 4 sulfate, heparin sulfate, dermatan sulfate and hyaluranan. People with mucopolysaccharide disease either do not produce enough one of the enzyme required to break down these sugar chain into simple molecule or they produce enzyme that don't work [2]. Morquio disease (MPS IV) is caused by a deficiency of N-acetylgalactosamine-6-sulfatase (MPS IV-A) or of β -galactosidase (MPS IV-B). Both result in the defective degradation of keratan sulfate [2,3]. The incidence of MPS is between 3.5 to 4.5/100000. The most common subtype is MPS III. Our case is a 8 month old son of a non-consanguineous parents from Sandwip, Chittagong, admitted to the Department of Paediatrics, Chittagong Medical College Hospital with a history of gibbus at thoracolumbar region for last two months. But on examination he was found to be macrocephalic, stunted and wasted. Skeletal survey had shown some consistent findings of Morquio disease and his urine for Mucopolysaccharidoses screening test is also positive. We have reported this case of MPS as it is rarer and among the MPS, Morquio disease is also not a commoner one [4,5]. It is also necessary for early differentiation of MPS IV from some treatable clinical entity like congenital hypothyroidism.

Key words

Morquio; mucopolysaccharidoses; skeletal survey.

Case

A 8 month old boy of non-consanguineous parents from Sandwip, Chittagong, admitted to the Department of Paediatrics, Chittagong Medical College Hospital (CMCH) with gibbus at thoracolumbar region for two months. His prenatal, natal and postnatal history was uneventful. He was on exclusive Breast Feeding for full six months along with right time introduction of complementary feeding with family diet. His developmental history was age appropriate.

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He had no history of repeated respiratory tract-ear infection, prolonged fever, trauma or had no history of contact with tuberculous Patient. His bowels and bladder habit is normal. His weight is 6.6 kg, length is 65cm, length for age(stunting)<-4SD and weight for length(wasting)<-1SD. Head is macrocephalic (OFC 49cm) having frontal bossing with wide open anterior fontanelle, posterior fontanelles is closed. Upper segment lower segment ratio is 1.7: 1. Arm span minus length is -5cm. His development is age appropriate.

Hands are stout, fingers are stubby but no Rhizomelic shortening is there. There is a 6 cm in length non erythematous, normal warmth, non tender angulation in the thoracolumbar region with no paravertebral swelling. He had no organomegaly. His ophthalmological examination reveals no abnormal finding and hearing is intact. There is no neurological abnormality. He has no mental retardation. Cardiorespiratory, gastrointestinal and all others systems reveal no abnormality. His thyroid hormone status is normal. (T_4 -1.21ng/dl and TSH-2.03IU/ml). Complete blood count, PBF and Urine R/M/E investigations results are normal. Roentherographic features in skeletal X-ray, had shown some consistent findings of Morquio disease. His urine for Mucopolysaccharidoses screening test is also positive.



Fig 1 : X-ray shows evidence of ant. beaking of upper lumbar vertebral bodies and kyphosis, expanded anterior end of ribs, squared iliac wing and constricted sacro-sciatic notches.



Fig 2 : Squared iliac wing, constricted sacro–sciatic notches, hypoplasia of femoral head epiphysis, shortened long bones :Features suggestive of Morquio disease.



Fig 3 : X-ray of both wrist joints(A/P) shows evidence of shortening of radius,Ulna and metacarpal bones along with abnormal flaring of distal end of radius and Ulna- some consistent findings of MPS IV.

From History , clinical features and X-ray and lab. investigation we diagnosed the case as Mucopolysaccharidoses IV (Morquio Disease).

Discussion

MPS is a group of inherited disease which is characterized by defective lysosomal enzymes which are responsible for the degradation of mucopolysaccharides which leads to the accumulation of incompletely degraded mucopolysaccharides in the lysosomes that affect various organs of the human body [3,4].

Morquio disease also known as mucopolysaccharidoses type IV, is an autosomal recessive disorder caused by deficiency of N-acetylgalactosamine-6-sulfate. Glycosaminoglycans is a long chain complex carbohydrate composed of uronic acid, amino sugars and neutral sugars. The mucopolysaccharidoses are part of the lysosomal storage disease family a group of 40 genetic disorders that when a specific organelles in our bodies lysosomal malfunction [6].

Both types of Morquio disease are characterized by short trunk and dwarfism, fine corneal deposition, skeletal dysplasia that are distinct from other MPS . Our patient had many skeletal deformities like kyphosis, gibbus, stout hands & stubby fingers.He had also macrocephaly and short stature. But this patient was presented with a normal cornea.

Normal intelligence, dwarfism, vulvular heart disease and hypermobility of the joint are the other features of this syndrome [7,8]. Though these are normal in our case. In our patient milestone of development was age appropriate.

Bone changes in this condition is diagnostic and in this case we also found many characteristic X-ray findings in skeletal survey.

Morquio disease is characterized by presence of keratosulfate in urine and Reily body in cytoplasm of leukocyte [6]. In this patient urine for Mucopolysaccharidoses screening test is also positive.

The commonest cause of death which usually occur around the 3rd and 4th decade is due to corpulmonale, valvular heart diseases [4]. Though in this case cardiovascular system examination revealed no abnormality.

There is no specific treatment for Morquio disease. Management is inevitably multidisciplinary and the paediatrician should play a lead role in the coordination of services for affected patients. Now enzyme replacement is a new horizon in the field of this case management. Symptoms can be treated accordingly. A spinal fusion may prevent permanent spinal cord injury in persons whose neck bone are underdeveloped and can be life saving [9-10].

Conclusion

Mucopolysaccharidoses IV (Morquio Disease) is a rarely found case. Early diagnosis based on clinical and roentgenographic findings is vital to differentiate it from some treatable paediatric cases like congenital hypothyroidism. Multidisciplinary approach is the backbone of this case management but now enzyme replacement is a new horizon in this field.

Disclosure

All the authors declare no competing interest.

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