MUCOPOLYSACCHARIDOSIS, A RARE METABOLIC DISORDER: A CASE REPORT

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Summary

Mucopolysaccharidosis (MPS) is a type of storage disorders which can affect the skin, liver, spleen, eye, brain and bone. Some variant present in early infancy and others present in late childhood. Clinical, biochemical and radiological evaluations are needed for a conclusive diagnosis. We present a case of MPS with various radiological findings of spines and skull which helped in the diagnosis of the condition in a resource poor setting like that of us.

Key words

Mucopolysaccharidosis (MPS); Glycoseaminoglycan (GAG); Hunter's syndrome.

Introduction

Mucopolysaccharidosis (MPS) is a broad spectrum of disorders caused by deficiency of one of a group of enzymes that degrades heparin sulphate, dermatan sulphate and keratin sulphate. The incidence is around 0.04-0.3% of the newborn and 1.5% of all congenital disorders(1). All mucopolysaccharidoses are autosomal recessive disorders, except for Hunter's syndrome (X-linked recessive). Patients suffering from MPS, usually, don't show characteristic clinical features from their birth which they develop later in their life.. Coarse facies, corneal clouding, hepatosplenomegaly, joint stiffness, hernias, skeletal abnormalities and mucopolysaccharides excretion in the urine, metachromatic staining in peripheral leukocytes and bone marrow are some general and special clinical findings. The average survival of the patient is around 20-30 years, and death is due to cardiac failure and infections to the gastrointestinal tract or to instability of atlantoaxial joint. Here we are presenting a case of MPS as it is rarely diagnosed in our setting and it has some interesting radiological features.

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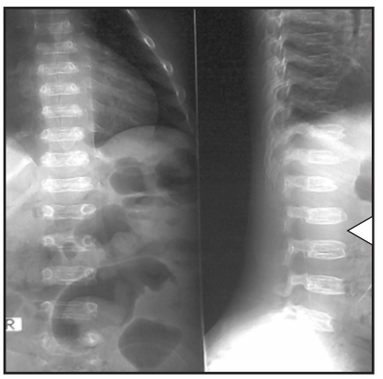
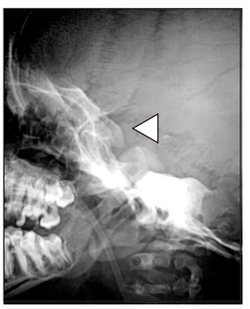
Case

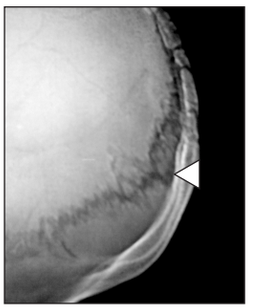
A 11-year male child attended outpatient department of Chittagong Medical College Hospital two years ago with progressive coarsening of facial features with macroglosia, short stature and difficulty in walking. He had joint stiffness and moderate hepatosplenomegaly. The boy was born normal vaginally, uneventfully and his mother gave no history of consanguinity of marriage. She noticed that milestone of development of the boy was a little delayed and progressively he was facing difficulty in going school or reading books or walking. He had no deafness but his IQ was found low in relation to his age. Skin findings were hypertrichosis, thickened skin, and multiple cobblestone skin lesions. He had mild respiratory distress on exertion. Thyroid function was found normal. Radiological survey of spine showed disc space widening, tongue shaped protrusion with beaking of vertebral bodies (Photograph 1). X ray skull with sella showed J shaped widening and separated cranial sutures(Photograph 2a, 2b). Based on characteristic clinical and radiological features MPS was diagnosed.



Photograph 1: Radiograph of thoracolumber spine- tongue shaped protrusion with beaking of vertebral bodies



Photograph 2a:
X-ray Skull, sella- J shaped



Photograph 2b:
X-ray skull, sutures- widened

Discussion

MPS was described first by two eminent scientist G. Hurler and C. Hunter [1]. Canadian internist Hunter, described the Hunter syndrome in London. Binswanger and Ullrich, described the different radiological changes in skeleton which are common in MPS. Among the different radiological findings , J-shaped sella with a enlarged skull, changes in the bodies of thoracic and lumber vertebrae (beaking of the body and widening of intervertebral space), pelvic bone hypoplasia, coxa valga, oar-shaped ribs (vertebral narrowing with anterior widening), expansion of metaphysis of long bones are the most common among the patients of MPS and these could be used as a diagnostic tool in a resource poor setting [2].

MPSs' involve multiple systems of the body and its course is progressive in nature. Clinical features, radiological abnormalities and laboratory findings are most often common in different varieties of MPS. Coarse facial features, enlarged liver, presence of glycoseaminoglycan (GAG) fragments in urine and leucocyte inclusion bodies are some common clinical and laboratory findings. Male are mainly affected by Hunter syndrome than female and it can be distinguished from other MPSs by its X-linked transmission pattern. Also, corneal opacity is not seen in Hunter syndrome but common in Hurler syndrome so it is said that "Hunter can Hunt [3,4]".

There are no curative treatment for MPS. But a variety of treatment options becoming available to improve the quality of life of the MPS patients. Among the most recent treatment options, affected enzyme replacement therapy (ERT) or allogenic bone marrow transplantation (BMT) are available in developed countries [5]. Outcome of treatment depends on the type of MPS, age at onset of disease, extent of clinical involvement and the pattern of donor genotype in case of BMT. In developed countries screening of the newborn just after birth for these disorders is being developed to avoid irreversible organ damage. Gene therapy is a promising modality of treatment but still experimental and yet to go a long way to get its full benefit [6].

Here the case was diagnosed as a case of Hunter syndrome when he had most of the features of MPS without corneal clouding. He was assessed further abroad where presence of GAG was confirmed in urine by ELISA. Due to financial constrain they could not have the regular follow up and one year later the boy died due to severe sepsis.

Acknowledgement

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Disclosure

All the authors declared no competing interest.

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