Morquio Disease with CNS Involvement

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Abstract

Morquio syndrome is a rare type of mucopolysaccharidosis. Our patient presented with uncontrolled seizures and gross skeletal deformity. He was suspected to be suffering from mucopolysaccharidosis based on his disease presentation but the diagnosis could be made as Morquio syndrome on the basis of the presence of keratan sulphate in the urine.

Key words: Morquio Disease, Mucoplysaccharidosis, Keratan sulphate

Introduction

Morquio syndrome was first described in 1929 by Morquio[1] and Brasilford[2] independently as cases of disorder characterized by short trunk, pectus carinatum and genu valga with normal intelligence. In 1960 this disorder was recognized as mucopolysaccharidosis caused by lysosomal accumulation and urinary excretion of the glycosaminoglycans (keratin sulphate)[3].

In type A Morquio there is deficiency of the enzyme N-acetyl-galactosamine -6-sulphatase and in type B there is deficiency of the enzyme betaglucoronidase[4]. In this disease keratin sulphate accumulates in connective tissue leading to severe somatic manifestations like seketal deformity and extraskeletal involvement. Risks are associated with cervical myelopathy, restrictive pulmonary disease and cardiac disease but there is generally no CNS involvement.

Morquio disease is a very rare type of mucopolysaccharidosis. The incidence is 1in 75,000[5].

The Case

8 year old boy born of nonconsanguinous marriage presented in paediatric emergency with active convulsion which lasted for 30 minutes and controlled by giving multiple i.v. anticonvulsants. History in details revealed that the boy was apparently well till 7 years of age except few episodes of hospitalization due to recurrent respiratory tract infections. Birth history was within normal limit.

Child had achieved the appropriate developmental milestones till 7 years. This was followed by difficulty in walking with frequent falls which gradually progressed to the extremity of inability to stand. There was marked deterioration in scholastic performances.

General survey revealed coarse facial features, pectus carinatum, multiple decayed teeth, kyphoscoliosis, short trunk, ulnar deviation of both the forearms.

Comprehensive nervous system examination revealed motor weakness and waddling of the gait.

Laboratory tests for detecting the altered biochemical parameters in blood provocative of seizures were found to be normal.

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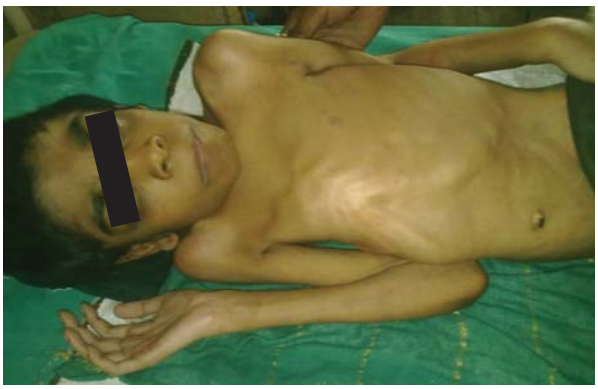

Fig 1: Showing coarse facial features, pectus carinatum, kyphoscoliosis.

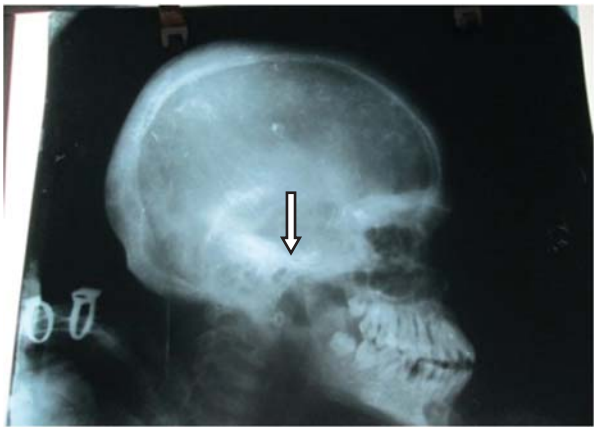

Fig 2: Thickened calvarium and J shaped sella.

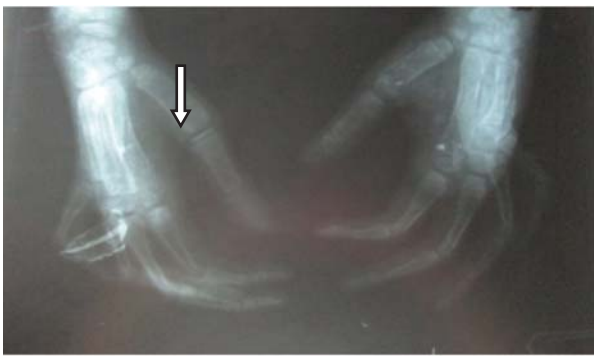

Fig 3: Bullet shaped phalanges

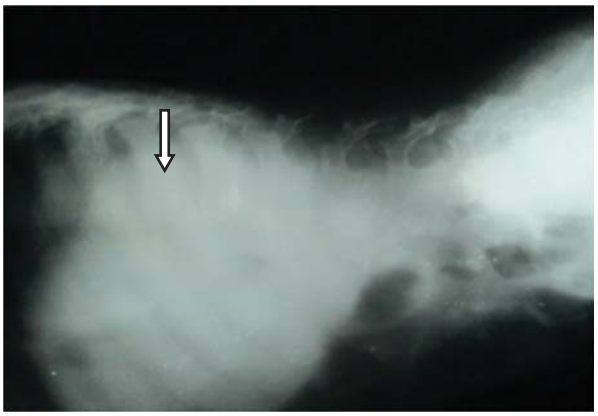

Fig 4: Spatular Vertebral body

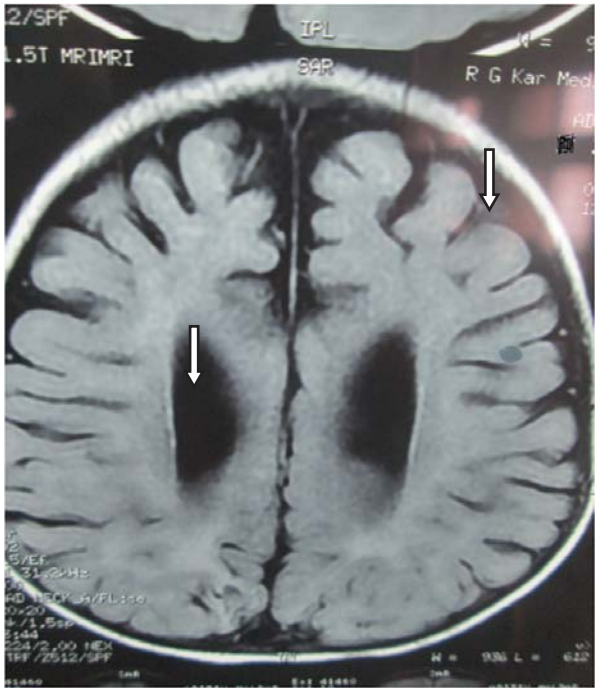

Fig 5: MRI brain- atrophy with ventriculomegaly.

X-Ray findings showed 1) thickened calvarium with J shaped sella 2) bullet shaped phalanges 3) spatular vertebral body.

The child was suspected to be suffering from mucopolysaccharidosis after conglomerating his clinical background, physical features with radiological findings.

Considering the above ophthalmological examination was done. Slit lamp examination of eyes revealed slight cloudiness of cornea with deposition of small dust like opacities. For further confirmation of the diagnosis chromatographic analysis of urine was done which showed increased amount keratin sulphate clinching to our ultimate diagnosis of MPS type IV commonly known as Morquio syndrome.

The child was treated symptomatically with anticonvulsants to control the seizures and antibiotic for his chest infections. The parents were explained about the nature of his disease and advised to visit the health workers to overcome his physical limitations.

Discussion

The Morquio syndrome is inherited metabolic disorder transmitted as autosomal recessive pattern. The patients suffering from this disorder appears healthy at birth and have normal intellectual growth. The common presentation is skeletal deformity and growth retardation beginning in the second and third

years of life. The child in our case simulated the same clinical picture except for the presence of severe neurological sequalae which generally is not familiar with Morquio syndrome. However skeletal deformity with CNS manifestations can be found in Hurler's syndrome (type I MPS)

The correct diagnosis was reached with a simple biochemical test of urinary detection of increased keratin sulphate. The novelty of our case lies in manifestation of CNS impairment in Morquio syndrome which is very rare.

Conclusion

Morquio syndrome presents with multiple complications like breathing problems, recurrent chest infections, spinal cord damage, vision problem and walking problems thus necessating multidisciplinary approach. Morbidity is high around 3rd to 4th decades. Through this case we emphasize the occurrence of disease with serious CNS manifestations and advocate genetic counselling.

We also highlight the importance of simple chromatographic analysis of urine in making right diagnosis. This test is very rapid, inexpensive and can be effectively carried out in areas of limited infrastructure.

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