ORIGINAL ARTICLE

Clinical and Radiological Diagnosis of Hurler–Scheie Syndrome

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ABSTRACT

Background: Doctors find difficulties in diagnosing rare syndrome as it is difficult to acquire adequate knowledge and experience of hundreds or thousands of rare conditions. Diagnosing rare inherited metabolic disorders such as mucopolysaccharidoses are also complicated by the lack of confirmatory laboratory tests in many areas of the world including Iraq. In the more developed countries, the diagnosis of mucopolysaccharidoses depends on urine tests for excessive mucopolysaccharides and enzyme assays. However, these tests are not available universally in all countries, and the diagnosis has to be made on clinical and radiologic findings. **Materials and Methods:** The clinical and radiological diagnosis of an Iraqi patient with Hurler–Scheie syndrome is presented. **Results:** A six years old girl with coarse facial features, stiff joints with contractures, normal mental and motor development, and normal neurological examination, and hearing impairment. Bone radiographs showed evidence of severe skeletal dysplasia with thickened calvarium. The girl had cardiac involvement, but she didn't have hepatosplenomegaly. The clinical diagnosis of the girl's condition was Hurler-Scheie syndrome because the changes of mucopolysaccharidosis were recognizable before two years with the absence of mental retardation and hepatosplenomegaly, and she didn't have the distinctive skeletal feature of Morquio syndrome. **Conclusion:** A confident clinical diagnosis of rare metabolic syndromes such as mucopolysaccharidoses necessitates remarkable clinical skills and vast experience due to the similarity between different types of mucopolysaccharidoses.

Key words: Diagnostic challenge, Hurler–Scheie syndrome, Iraq

INTRODUCTION

Doctors find difficulties in diagnosing rare syndrome as it is difficult to acquire adequate knowledge and experience of hundreds or thousands of rare conditions. Diagnosing rare inherited metabolic disorders such as mucopolysaccharidoses are also complicated by the lack of confirmatory laboratory tests in many areas of the world including Iraq. In the more developed countries, the diagnosis of mucopolysaccharidoses depends on urine tests for excessive mucopolysaccharides and enzyme assays. However, these tests are not available universally in all countries, and the diagnosis has to be made on clinical and radiologic findings.[1-3]

MATERIALS AND METHODS

The clinical and radiological diagnosis of an Iraqi patient with Hurler–Scheie syndrome is presented.

RESULTS

A 6-year-old girl experiencing easy fatigability, exceptional dyspnea, and difficulty in walking was seen at the Children Teaching Hospital of Baghdad Medical City. She had coarse facial features including thick eyebrows, a flat nasal bridge, thick lips, and enlarged mouth. She did not have the characteristic chest deformity of Morquio syndrome [Figure 1]. The child also had very stiff joints with contractures

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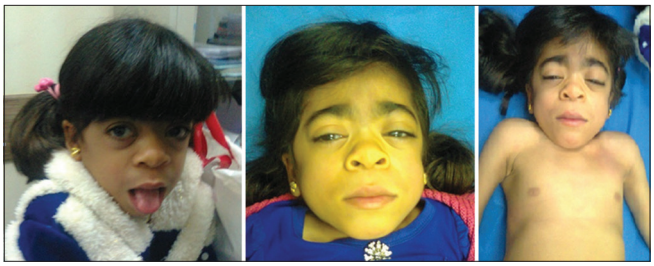

Figure 1: The girl had coarse facial features including thick eyebrows, a flat nasal bridge, thick lips, and enlarged mouth. She did not have the characteristic chest deformity of Morquio syndrome

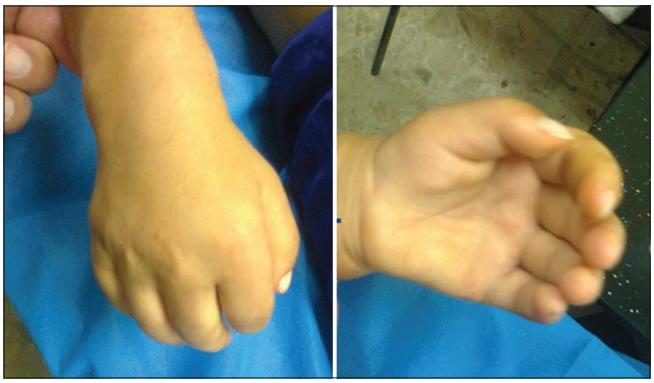

Figure 2: The girl had very stiff joints with contractures hands

at the knees and hands [Figure 2], and she was walking with some difficulty. Her height was 97 cm. Her mental and motor developments were normal. Neurological examination revealed normal findings with no evidence of spasticity, and she had no hearing impairment. Bone radiographs [Figure 3] showed evidence of severe skeletal dysplasia with bone enlargement, irregular shape, and bowing of bones. Skull radiographs [Figure 4] showed thickened calvarium.

The skeletal abnormalities in the girl were not characteristic of Morquio syndrome which is not associated with a significant coarsening feature as in this girl. She did not have hepatosplenomegaly. Echocardiography showed mild mitral and aortic regurgitation. Parents were relatives and healthy. She had two normal sisters aged 13 and 16 years, respectively. However, she had two older brothers having the same condition. The brothers aged 11 and 20 years, respectively. Both brothers were very short and were reported to have cardiomegaly and heart disorder. The parents believed that their affected children were normal at birth and were normal during the first 2 years of life.

The clinical diagnosis of the girl's condition was Hurler–Scheie syndrome because the changes of mucopolysaccharidosis were recognizable before 2 years with the absence of mental retardation and hepatosplenomegaly, and she did not have the distinctive skeletal feature of Morquio syndrome.

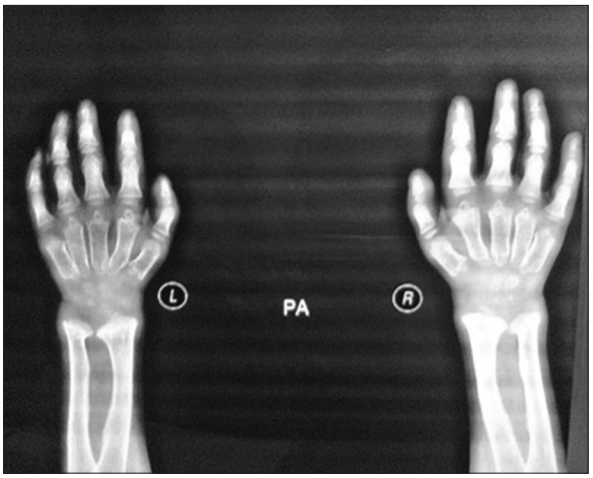

Figure 3: Bone radiographs of the patient showed evidence of severe skeletal dysplasia with bone enlargement, irregular shape, and bowing of bones

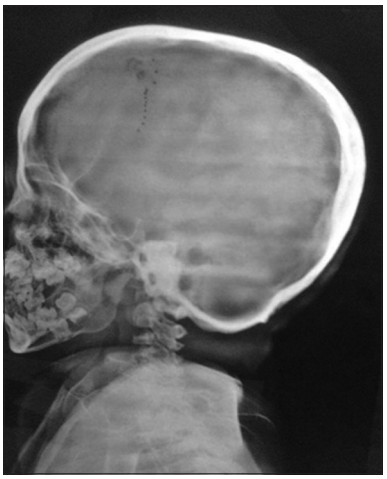

Figure 4: Skull radiographs showing thickened calvarium

DISCUSSION

Mucopolysaccharidoses syndromes were first described by Gertrud Hurler (1889–1965), a German pediatrician in 1917, and by Charles A Hunter (1873–1955) in 1919 as syndromes of chondrodystrophic changes in the skeleton, corneal opacities, hepatosplenomegaly, and mental retardation. In 1936, Ellis *et al.* called the mucopolysaccharidoses syndrome gargoylism.[3-6]

Hurler syndrome is the severe form of the disorder with death usually occurs by 10 years of age that the syndrome is characterized by coarse facial features, short stature, skeletal deformities, joint stiffness, and mental retardation with onset of disease usually between 2 and 4 years of age. Scheie syndrome is the mild form of mucopolysaccharidoses. The onset of significant symptoms is usually recognized after the age of 5 years with diagnosis generally made after 10 years. Scheie syndrome is associated with joint stiffness, aortic valve disease, corneal clouding, retinal degeneration, and

obstructive airway disease. However, patients with Scheie syndrome have normal intelligence and significant mental retardation does not occur.

Hurler–Scheie syndrome is a clinical phenotype that is intermediate between Hurler and Scheie syndromes. The physical involvement is progressive but with little or no intellectual dysfunction. Symptoms are usually observed between 3 and 8 years, and survival to adulthood is common. Cardiac involvement and upper airway obstruction contribute to clinical mortality.[3,7-10]

Morquio syndrome is associated with a distinctive skeletal dysplasia, short trunk and neck, fine corneal deposits, and the absence of mental retardation. The appearance of genu valgus and waddling gait with a tendency to fall is early symptoms of Morquio syndrome. Coarsening of facial features is less significant than in other types of mucopolysaccharidoses.[3]

Hurler syndrome and Hunter syndrome are associated with significant mental retardation and cannot be considered in the differential diagnosis of a girl with mucopolysaccharidosis and normal intelligence. Hunter syndrome has X-linked recessive inheritance and occurs in males.[3-6]

Coarsening of facial features and hepatomegaly is less significant in Morquio syndrome than in other types of mucopolysaccharidosis. Morquio syndrome, Scheie syndrome, and Hurler–Scheie syndrome may occur in girls without significant mental retardation, but they are not associated with early onset and are not diagnosed before the age of 2 years.[3-10]

Maroteaux–Lamy syndrome is associated with early onset of symptoms and the diagnosis can be made before the age of 2. It is characterized by coarse facial features, joint stiffness, valvular heart disease, and dysostosis multiplex, and the absence of mental retardation. Mild and intermediate forms of Maroteaux–Lamy syndrome are very similar to Scheie syndrome.[3,11-13]

The emergence of the new enzyme replacement therapies for some types of mucopolysaccharidoses raises the importance of the clinical diagnosis of such disorders to give the patients the chance to have the new therapy in another country when this is possible.[3]

CONCLUSION

A confident clinical diagnosis of rare metabolic syndromes such as mucopolysaccharidoses necessitates remarkable clinical skills and vast experience due to the similarity between different types of mucopolysaccharidoses.

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