

WORLD JOURNAL OF NUTRITION AND METABOLISM, DIETETICS

Clinical and Radiologic Diagnosis of Maroteaux-Lamy Syndrome

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Article Information

Article Type:	Research	*Corresponding author:	Citation: Al-Mosawi AJ (2020) Clinical and Radiologic Diagnosis of Maroteaux-Lamy Syndrome. World J Nutr Metabol Diet, 1(1);1-3
Journal Type:	Open Access	Aamir Jalal Al Mosawi	
Volume:	1 Issue: 1	Advisor in Pediatrics and Pediatric Psychiatry Children Teaching Hospital of Baghdad Medical City, Head	
Manuscript ID:	WJNMD-1-102	Iraq Headquarter of Copernicus Scientists International Panel Baghdad	
Publisher:	Science World Publishing	Iraq	
Received Date:	24 December 2019	Email: almosawiaj@yahoo.com	
Accepted Date:	25 January 2020		
Published Date:	26 January 2020		

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ABSTRACT

Background: Diagnosis of rare diseases or disorders is understandably challenging because it is unfeasible for practicing physicians to make themselves familiar with hundreds of rare disease. The diagnosis of rare inherited metabolic syndromes such as mucopolysaccharidoses is additionally complicated by the lack of confirmatory sophisticated laboratory tests in many areas of the world. In the more developed countries, the diagnosis of mucopolysaccharidoses depends on urine tests for excessive mucopolysaccharides and enzyme assays. However, these tests are not easily accessible in countries like Iraq, and the diagnosis has to rely on clinical and radiological findings.

Materials and Methods: The clinical and radiologic diagnosis of an Iraqi patient with Maroteaux-Lamy syndrome is described.

Results: The clinical diagnosis of this girl with early onset mucopolysaccharidosis and the abnormalities were recognizable before the age of two. The clinical a radiologic diagnosis was Maroteaux Lamy syndrome because of the absence of mental retardation and the presence of hepatosplenomegaly.

Conclusion: Clinical diagnosis of rare metabolic syndromes like mucopolysaccharidoses requires magnificent clinical skills and huge experience because of the similarity between various types of mucopolysaccharidoses.

KEYWORDS

Maroteaux Lamy syndrome, Diagnostic challenge, Iraq

MATERIALS AND METHODS

The clinical and radiological diagnosis of an Iraqi patient with Maroteaux-Lamy syndrome is described.

RESULTS

Five and half years old girl who is known to have a metabolic storage disease was seen during April, 2016 because of having breathing problem that is mostly attributed to recurrent respiratory infections and obstructive airway disease. Her parents were relatives and her older sister had the same condition and died before the age of 10 years from cardiac problem and she was blind before death, but she didn't experience intellectual deterioration.

At the age of one and half year (April, 2012), it was clear that this patient had coarse facial features and abdominal enlargement. Her motor and mental development was considered normal during that time.

Metabolic screening tests were performed and included urinary tests for homocystine, cystine, phenylketonuria, homogentistic acid, and ketoacids, they were all negative. There was no glucosuria and Benedict's test for reducing substances was also negative. Urine chromatography for amino acids showed normal findings, but serum chromatography was positive for methionine.

During April, 2012, her heart was also evaluated with echocardiography which showed persistent thymus which may give the impression of cardiomegaly. Figure-1 shows the coarse facial features of the child at the age of five and half years, during April, 2016, and shows the abdominal enlargement caused by hepatosplenomegaly with spleen reaching the umbilicus. The child had thick eyebrows, a depressed nasal bridge, thick lips, and larged mouth and tongue (macroglossia). The child also had very stiff joints with contractures at the knees, and her walking was rather difficult. Her mental and motor development was normal. Neurologic examination was normal with no evidence of spasticity, and she has no hearing impairment.



Figure 1: The girl had coarse facial features and abdominal enlargement caused by hepatosplenomegaly with spleen reaching the umbilicus.

Bone radiographs showed evidence of severe skeletal dysplasia with bone enlargement, irregular shape, and bowing of bones (Figure 2).

The clinical diagnosis of this girl with early onset mucopolysaccharidosis and the abnormalities were recognizable before the age of two. The clinical and radiologic diagnosis was Maroteaux Lamy syndrome because of the absence of mental retardation and the presence of hepatosplenomegaly.

DISCUSSION

Mucopolysaccharidosis 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 and colleagues called the Mucopolysaccharidosis syndromes gargolism [3-6].

Hurler syndrome is the severe form of the disorder with death usually occurs by 10 years of age. 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 yr of age. Scheie syndrome is the mild form of Mucopolysaccharidosis. Onset of significant symptoms is usually recognized at about the age of six years with diagnosis generally made after ten years. Scheie Syndrome is associated with stiff joints, aortic valve abnormalities, corneal clouding, degeneration of the retina and airway obstruction. However, patients with Scheie syndrome have normal intelligence and significant mental retardation doesn't occur.

Hurler-Scheie syndrome is a clinical phenotype that is intermediate between Hurler and Scheie syndromes. The somatic involvement is progressive, but with little or no intellectual dysfunction. Symptoms is commonly observed between three and eight years of age, 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 skeletal dysplasia that is distinct from other mucopolysaccharidosis with a short trunk and



Figure 2: Bone radiographs showing evidence of severe skeletal dysplasia with bone enlargement, irregular shape, and bowing of bones

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 and Hunter syndromes are characterized by significant mental retardation and cannot be included in the differential diagnosis of a girl with mucopolysaccharidosis without significant mental retardation. Hunter syndrome has and X-linked recessive inheritance, and occur in males [3-6].

Coarsening of facial features and hepatomegaly are less significant in Morquio syndrome than in other types of mucopolysaccharidosis. Morquio syndrome can not be considered in the differential diagnosis of a girl with mucopolysaccharidosis with hepatosplenomegaly and significant coarsening of facial features. Scheie and Hurler-Scheie syndromes can occur in girls without significant mental retardation but they are not associated with early onset and are generally not diagnosed before the age of two years [3-10].

Maroteaux Lamy syndrome commonly has early onset of symptoms and the diagnosis earlier than the age of two. It is characterized by coarse facial features, joint stiffness, valvular heart abnormality, and dysostosis multiplex, and the lack of mental retardation. Mild and intermediate forms of Maroteaux Lamy syndrome are very similar to Scheie syndrome [3,11,12,13].

Maroteaux-Lamy syndrome was also called Polydystrophic dwarfism. Mucopolysaccharidosis VI (MPS6) and Arylsulfatase B deficiency (ARSB deficiency). It is an autosomal recessive type of mucopolysaccharidosis which is a lysosomal storage disorder that was first described in 1963 by French physicians Pierre Maroteaux (1926-) and his mentor Maurice Emil Joseph Lamy. They described the condition as a novel dysostosis associated with increased urinary excretion of chondroitin sulfate.

Children with Maroteaux-Lamy syndrome are generally not mentally retarded, but have many of the physical symptoms of Hurler syndrome. Maroteaux-Lamy syndrome has a variable severity of manifestations include usually include short stature, hepatosplenomegaly, dysostosis multiplex, stiff joints, corneal clouding, cardiac abnormalities, and facial dysmorphism. Affected children will gradually develop a shortening of the trunk, crouched stance, restricted joint movement, and a protruding abdomen and forward- curving spine. The skeletal changes are progressive and restrict movement. Affected children may have umbilical hernia or inguinal hernias. Most children with this disorder develop cardiac abnormality mostly in the form of valvular dysfunction [3,11,12,13].

The recent availability of treatment with enzyme replacement for some types of mucopolysaccharidoses made the clinical diagnosis of such disorders of practical value as it may give the patients the chance to have the new treatment in an other country when this is possible [3].

CONCLUSION

Clinical diagnosis of rare metabolic syndromes like mucopolysaccharidoses requires magnificent clinical skills and huge experience because of the similarity between various types of mucopolysaccharidoses.

ACKNOWLEDGEMENT

The author would to express his gratitude for the parents of the patient who accepted publishing her photos.

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