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Echocardiographic manifestations of a 17-year old girl with Mucopolysaccharidoses type I (Hurler syndrome)

Hamide Khesali, MD; Niloufar Akbari Parsa, MD*

Corresponding Author: Niloufar Akbari Parsa, MD
Echocardiography Research Center, Rajaei Cardiovascular Medical and Research Center, Tehran, Iran.
Email: ni_pa85@yahoo.com

Abstract

Mucopolysaccharidoses are a group of rare genetic metabolic disorders with significant multi-organ failure. In this case report we present a case of mucopolysaccharidoses type I (Hurler syndrome) and discuss about the cardiac manifestations and echocardiographic features of the disease.

Keywords

Mucopolysaccharidoses; hurler syndrome; transthoracic echocardiography.

Introduction

Mucopolysaccharidoses (MPS) type I is a fatal genetic lysosomal storage condition. Deficiency of the lysosomal hydrolase IDUA leads to the glycosaminoglycans (GAG)accumulation within the CNS, lungs, heart, liver, cartilages and bones. Cardiac involvement is seen in 70%-80% of patients and is due to GAG deposition in the valves, coronary arteries, myocardium, and conduction system. Cardiovascular pathologies include mitral and aortic valves thickening resulted in mitral and aortic regurgitation, occlusion of the coronary arteries, endocardial thickening, and dilated cardiomyopathy, also stenosis of the renal arteries and abdominal aorta could happen. We present a case of MPS I (Hurler syndrome) with peripheral edema and dyspnea to underline the echocardiographic manifestations of the cardiac involvement.

Case Presentation

A 17-year old girl with MPS I (Hurler syndrome) diagnosed in early infancy presented to the emergency ward with dyspnea and progressive peripheral edema. She had thick skin and coarse facial features like a large head, broad nose, broad supraorbital ridges, periorbital edema, thick lips and enlarged tongue (Figure 1). She had visual problems and used high prescription strong glasses and also had auditory loss

and was candidate for ear surgery and hearing aid use. She suffered from skeletal deformities like short stature, thoracolumbar kyphosis, clawed hand and prominent hallux valgus in lower extremities (Figure 2). She showed severe restrictive lung disease pattern in her previous spirometry study. She was oriented and did not show cognitive impairments. She mentioned a history of heart failure based on her last cardiology visit. She had a cousin suffering from the same disease. She used furosemide, losartan, metoprolol succinate and Aldurazyme (Iaronidase). She had S3 and a holosystolic III/VI murmur in fifth intercostal space in auscultation. Electrocardiogram showed sinus rhythm with LBBB pattern and LVH and LV strain (Figure 3). Chest X-ray demonstrated huge cardiomegaly (Figure 4).

In transthoracic echocardiography she had severe left ventricle(LV) and right ventricle(RV) enlargement and a huge left atrium with a total deviation of interatrial septum to right. LV and RV functions were severely reduced with an LV ejection fraction:20%, LV walls had top normal thickness based on her BSA (body surface area). There was prominent thickening and prolapse of all valves resulted into severe mitral regurgitation and tricuspid regurgitation with mitral and tricuspid annular dilation; and moderate aortic regurgitation and up to moderate pulmonary regurgitation, there was no valvular stenosis, patient had a plethoric inferior vena cava and severe pulmonary hypertension (systolic pulmonary artery pressure: 70 mmHg, mean pulmonary artery pressure: 35-40 mmHg). There was no cardiac shunt. Small circumferential pericardial effusion was present (Figure 5; Supplementary video 1).

Discussion

The MPS consist of rare genetic metabolic disorders due to deficiencies or absence of the lysosomal enzymes effective on sequential degradation of GAGs and leads to accumulation of the substrate in different tissues result insignificant multi-organ failure. Patients affected by MPS might appear healthy at birth but show numerous clinical symptoms and signs like remarkable growth retardation, skeletal problems, coarse facial features, reduced joints range of motion, hearing loss, corneal clouding and visual problems, and cardiac involvement mostly valvular heart disease. Seven types of MPS (I, II, III, IV, VI, VII, and IX), caused by eleven enzymes deficiencies, have been characterized [1].

MPS I is caused by deficiency in a-L-iduronidase enzyme with autosomal recessive inheritance, resulted in large amount of urinary excretion of GAGs, mostly heparan sulfates and dermatan. Symptoms include atypical facial manifestations, normal cognition or developmental and mental retardation, short stature, dysostotic multiplex, stiffness of the joints, visual problems, respiratory failure and infections, valvular involvement and cardiomyopathy and hepatosplenomegaly. Patients affected by MPS I have less life expectancy [2].

Cardiac involvement in MPS I mostly include valvular heart disease and coronary arteries stenosis. Also, narrowing of the renal arteries leading to systemic hypertension and occlusion of the abdominal aorta has been described [3]. The most prevalently involved valves are the mitral and the aortic valves which are significantly thickened due to infiltration of GAG into the valves tissues and collagen excessive deposition [4]. Functional manifestations are restricted mobility of the valves, that causes regurgitation, and, to a lesser degree, valvular stenosis which might result in volume and pressure overload, ventricular dilation and/

or hypertrophy, and consequently to systolic and/or diastolic dysfunction [5,6]. Deposition of GAG in the vessels causes narrowing of the coronary arteries [4,7] which can be greater than 75%. However, it still remains unknown why the coronary arteries are mostly involved [4].

Transthoracic echocardiography is avaluable and safe diagnostic modality for evaluating ventricular and valvular involvement and function in MPS patients and should be used for diagnosis and follow up of all patients with the disease, to assess progression of cardiac abnormalities with age [2].

Enzyme replacement therapy (ERT) seems to be helpful in relieving cardiac manifestations and might show better outcomes when prescribed at younger age. Early prescription of ERT before the initiation of the irreversible cardiac injurycanlead to better clinical prognosis for the patients [1].

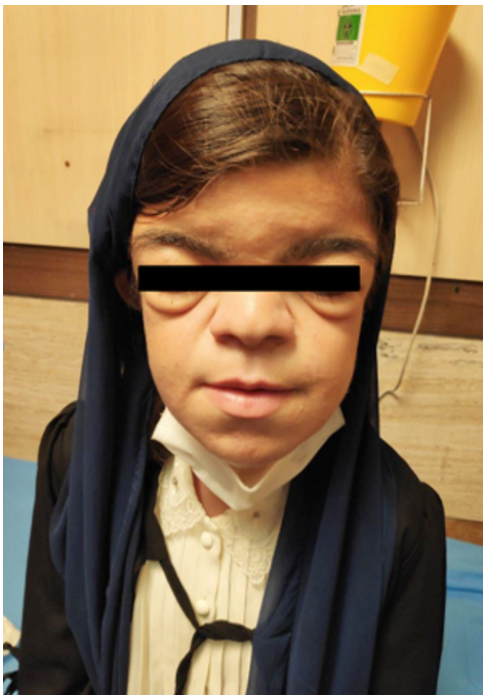

Figure 1: She had thick skin and coarse facial features like a large head, broad nose, broad supraorbital ridges, periorbital edema, thick lips and enlarged tongue.

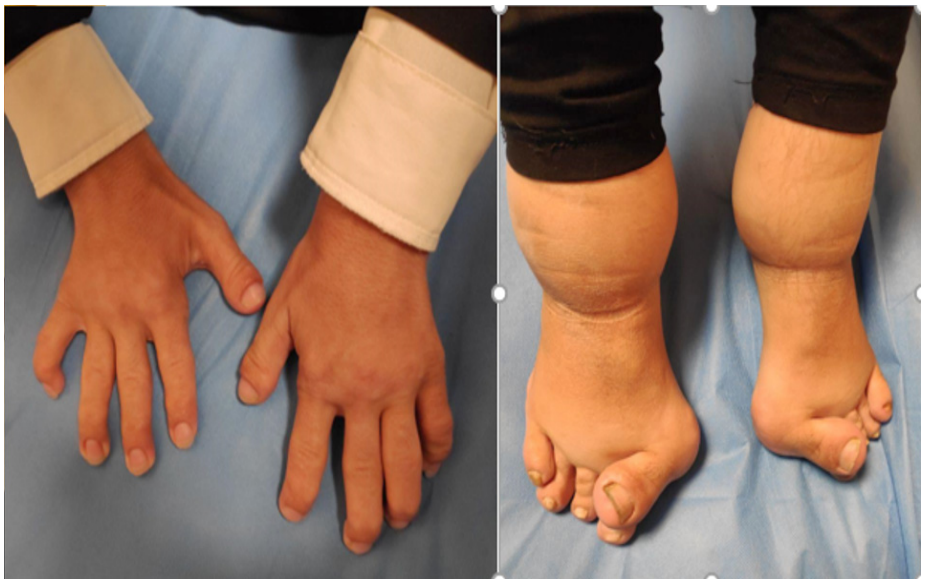

Figure 2: She had clawed hand and prominent hallux valgus in lower extremities.

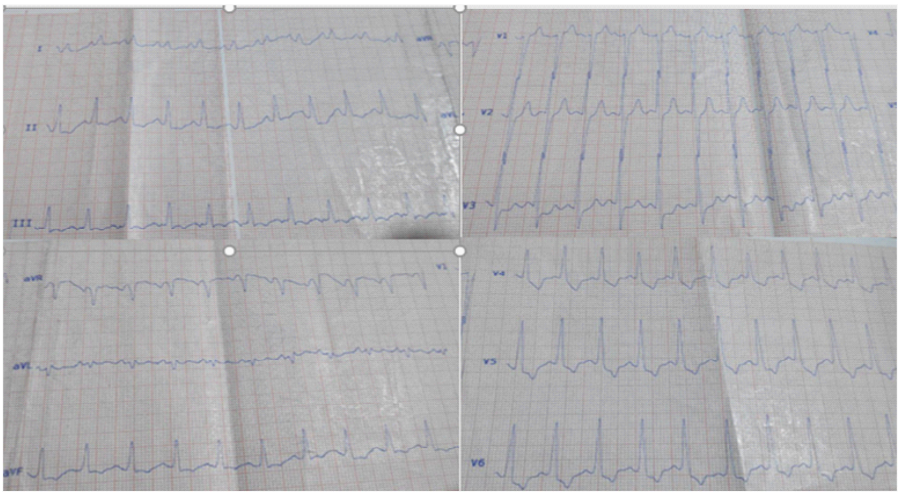

Figure 3: Electrocardiogram showed sinus rhythm with LBBB pattern and LVH and LV strain.

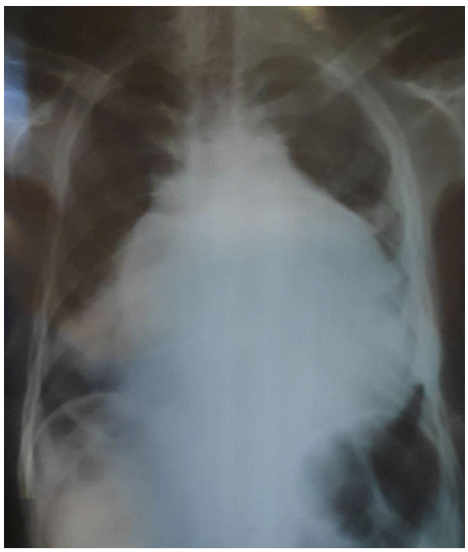

Figure 4: Chest X-ray demonstrated huge cardiomegaly.

Conclusion

Although MPS is a rare genetic disease, but the majority of the affected patients show cardiac problems. Early diagnosis of the disease and ERT can prevent some irreversible cardiac damage, so all physicians should be aware of features, symptoms and cardiac manifestations.

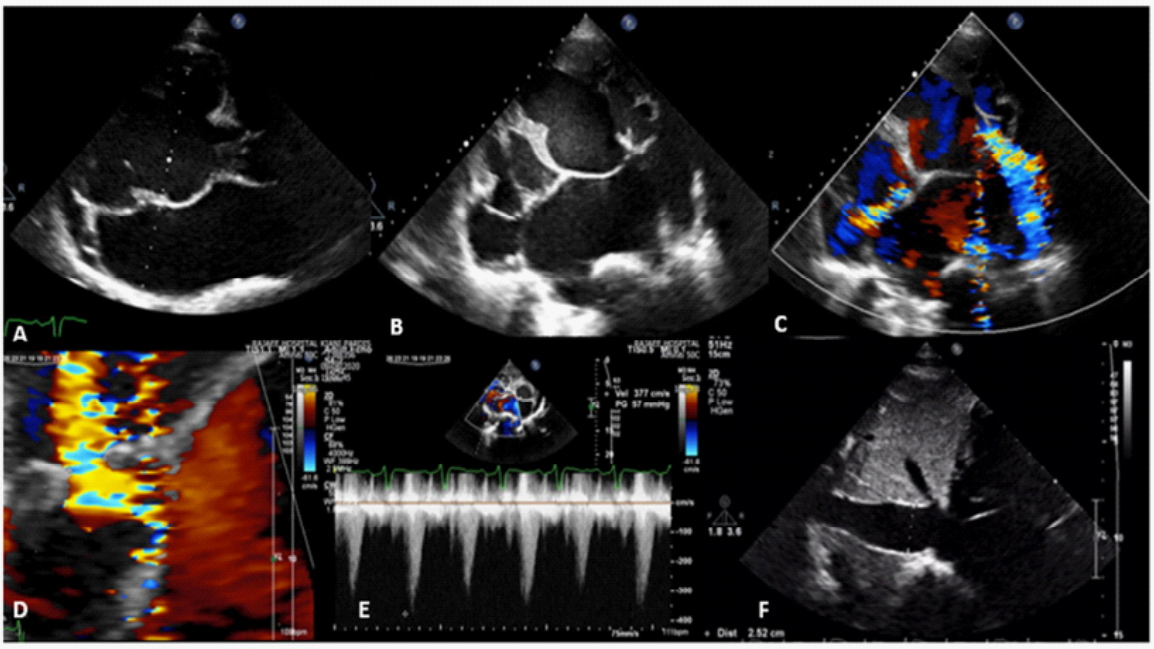

Figure 5: A,B: Severely enlarged both ventricles and left atrium with severe dysfunction and severely thickened and prolaptic mitral and tricuspid valves. **C,D:** Severe mitral and tricuspid regurgitations. **E,F:** Plethoric inferior vena cava and severe pulmonary hypertension.

References

1. HY. Lin, et al, Cardiac structure and function and effects of enzyme replacement therapy in patients with mucopolysaccharidoses I, II, IVA and VI, Mol. Genet. Metab. 2016.

2. Martins AM, Dualibi AP, Norato D, et al. Guidelines for the Management of Mucopolysaccharidosis Type I. The Journal of Pediatrics. 2009; 155(4): 32-46.

3. Taylor DB, Blaser SI, Burrows PE, et al. Arteriopathy and coarctation of the abdominal aorta in children with mucopolysaccharidosis: Imaging findings. AJR Am J Roentgenol. 1991; 157: 819–823.

4. Hampe CS, Eisengart JB, Lund TC, et al. Muccopolysaccharidosis Type I: A Review of the Natural History and Molecular Pathology. Cells. 2020; 9:1838.

5. Leal GN, De Paula AC, Leone C, Kim CA. Echocardiographic study of pediatrics patients with mucopolysaccharidosis. Cardiol. Young. 2010; 20: 254–261.

6. Krovetz LJ, Lorincz AE, Schiebler GL. Cardiovascular Manifestations of the Hurler Syndrome: Hemodynamic and Circulation 1965: 31; 132–141.

7. Braunlin EA, Hunter DW, Krivit W, Burke BA, Hesslein PS, Porter PT, Whitley CB. Evaluation of coronary artery disease in the Hurler syndrome by angiography. Am J Cardiol. 1992: 69; 1487–1489.

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Authors Information: Hamide Khesali, MD; Niloufar Akbari Parsa, MD*
Echocardiography Research Center, Rajaei Cardiovascular Medical and Research Center, Tehran, Iran.

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