Case Report: Aortic Stenosis in Mucopolysaccharidosis Type 1

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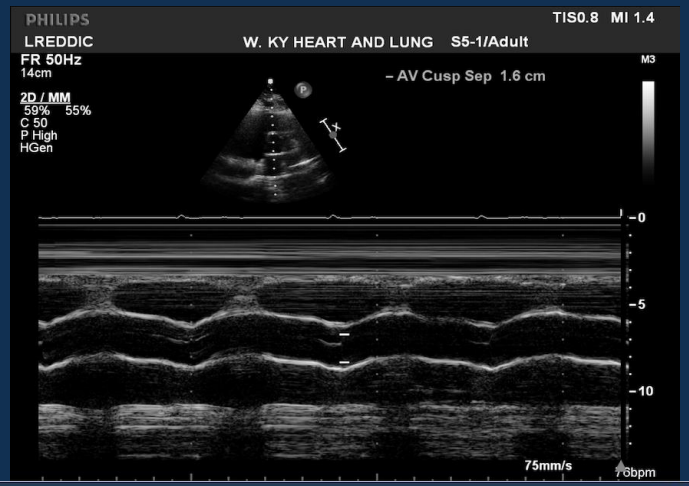
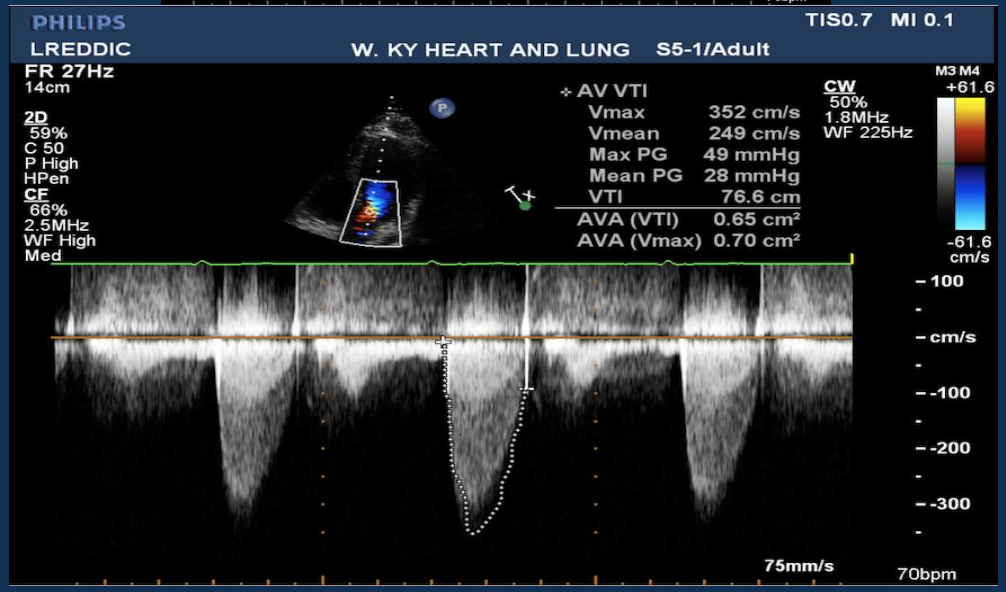
Introduction

- MPS I is an autosomal recessive lysosomal storage disorder that is caused by deficient enzyme activity of α-l-iduronidase (IDUA), leading to lysosomal accumulation of glycosaminoglycans (GAGs) in multiple tissues throughout the body including the heart.
- Mucopolysaccharidosis type I can be classified as three clinical sub-types; Hurler syndrome, Hurler-Scheie syndrome and Scheie syndrome, with the scale of severity being such that Hurler syndrome is the most severe and Scheie syndrome the least severe.
- Signs and symptoms include stiffened joints, skeletal problems, carpal tunnel syndrome, valvular abnormalities, upper airway Infections, obstructive sleep apnea, corneal clouding, spinal cord compression, enlarged liver and spleen, hearing loss, delayed cognitive development, coarse facial features, communicating hydrocephalus, and abnormally shaped teeth.

Case Presentation

- A 24-year-old Caucasian woman with PMH of migraine, anxiety disorder, vitamin-D deficiency and mucopolysaccharidosis type 1 (Hurler-Scheie) diagnosed in 2007.
- Symptoms started at 6 years of age, when she "froze up" and could not move or talk, with syncopal episodes.
- She presented to our clinic with sharp, pinpoint chest pain which is worse with palpation.
- Other signs and symptoms included corneal clouding with a 20/40 vision, joint/muscle/back pain, missing one rib, pars fracture in her lumbar spine, enlarged spleen and liver, slightly enlarged left kidney, tightness in calves which caused her to walk on toes, and headaches.
- On physical exam, grade 2/6 early systolic murmur was auscultated at RUSB.
- Transthoracic echocardiogram showed moderate valvular aortic stenosis and mild aortic regurgitation with borderline concentric left ventricular hypertrophy present.





Discussion

- Patients with MPS I may develop storage of GAGs within and around the mitral and aortic valve leaflets results in thickening and stiffening which can be progressive. Mitral regurgitation is the more common valvular disease in severe MPS I.
- Cardiac evaluation through serial monitoring of ventricular size, wall thickness and function at regular intervals at least every 2 years (according to Muenzer 2009) with echocardiography is useful in the treatment of patients.
- Cardiac evaluation should always include echocardiography to define the involvement of the valvular apparatus and its extent.
- Cardiological follow-up should be performed yearly for mild or stable valvular disease, and at least twice a year for patients with severe valvular disease and signs of suboptimal heart function.
- A surgical intervention for valvular replacement is therefore indicated in: a) symptomatic patients without significant comorbidities b) asymptomatic patients with severe valvular diseases and with evident sign of disease progression (systolic function impairment, chamber dilation, pulmonary hypertension, arrhythmias).

Conclusion

We learned about a rare case of aortic stenosis associated with MPS. Cardiological follow-up should be performed yearly for mild or stable valvular disease, and at least twice a year for patients with severe valvular disease with signs of suboptimal heart function.

References

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