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A Rare Case of Von Gierke causing Severe Aortic Stenosis

Srujana Dasari
HCA Healthcare, srujana.dasari@medicalcityhealth.com

Akhil R. Gade
HCA Healthcare, Akhil.gade@medicalcityhealth.com

Nethuja Salagundla
HCA Healthcare, nethuja.salagundla@medicalcityhealth.com

Chandralekha Ashangari
HCA Healthcare, Chandralekha.Ashangari@MedicalCityHealth.com

Rashonda Carlisle
HCA Healthcare, rashonda.Carlisle@medicalcityhealth.com

Von Gierke Aortic Stenosis

Srujana Dasari MD, Akhil Reddy Gade MD, Nethuja Salagundla MD, Chandralekha Ashangari MD, Rashonda Carlisle MD

Introduction

Von Gierke disease is an inherited Glycogen Storage disease - type 1 resulting from deficiencies in the specific enzymes glucose 6 phosphatase(1a), and/or glucose 6 phosphate translocase(1b) glycogen metabolism pathway. Von Gierke's disease is commonly seen in the pediatric population and is known for his hepatic and renal manifestations. Here we discuss a rare case of Von Gierke's disease causing some rare cardiac manifestations.

Case

41yo M with PMHx significant for Von Gierke type 1a, HFrEF complicated by VT s/p ICD, HTN who presented for complaints of continued SOB with new muscle soreness. On arrival, lactic acid was elevated at 5.2, and troponin and EKG were not obtained in the ED.

Von Gierke's Disease prevalence is one in 100,000

Most often seen in pediatric patients. Rare to see adults.

Usually hepatic and renal complication, cardiac complications are rare in this GSD.

Results

Trans-thoracic echocardiogram obtained was consistent with HFrEF with a LVEF 30-35%, severe diffuse hypokinesis, bileaflet aortic valve, severely calcified aortic stenosis with a peak systolic velocity 3.2m/sec, peak systolic gradient 50 mmhg, mean gradient 30 mmHg, and valve area 1.1. The patient was evaluated for aortic valve replacement. Left and right heart catheterization show an invasive gradient of 20 mmHg and angiographically normal epicardial coronary arteries.

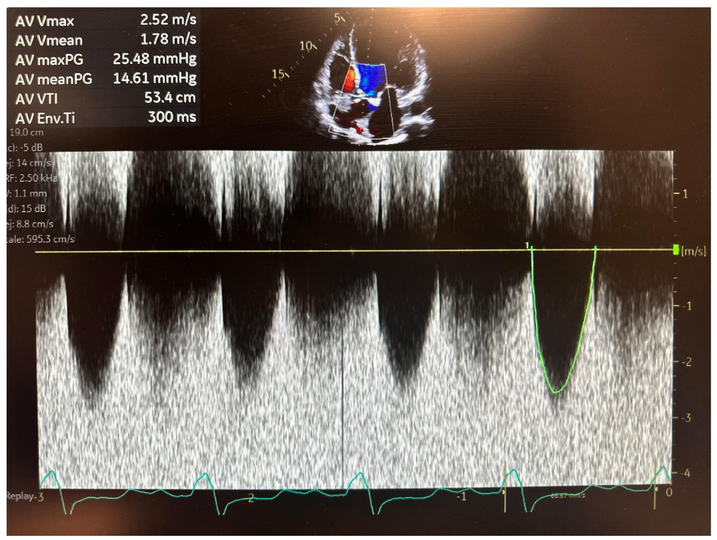

Figure 1: Echocardiogram with peak systolic velocity 3.2m/sec, peak systolic gradient 50 mmhg, mean gradient 30 mmHg,

Discussion

As mentioned, Von Gierkes is not typically seen in this middle-aged group let alone with unexplained cardiac manifestations. Our patient had heart failure for 10 years before presentation, and risk factors for CAD and ischemic cardiomyopathy were ruled out. Although not confirmed by biopsy, as the management would not change, the etiology is likely attributed to this patient's Von Gierke disease due to the deposition of glycogen in the cardiac membrane. This patient lived longer than most individuals with this disease. It is important to bring awareness to this rare disease with even more rare cardiac manifestations that have not been well studied. Patients should undergo screening from an early age to identify cardiac manifestations and be provided with appropriate resources including genetic counseling.

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