Atypical Presentation of Cardiac Fabry’s Disease
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LEARNING OBJECTIVES

- Recognize that Fabry’s disease (FD) is a potential etiology of acute coronary syndrome
- Have a high suspicion for myocardial infarction in patients with FD presenting with sudden onset chest pain.

CASE PRESENTATION

A 65-year-old woman with a history of FD managed with enzyme replacement therapy presented to the emergency department for evaluation of sudden onset heaviness in the center of her chest.

HPI: Central chest pain, S10 intensity, radiating down both arms associated with shortness of breath, diaphoresis and lightheadedness. First episode of similar symptoms. Significant family history of FD in her father who suffered from an unspecified cardiac disease. Patient recently had a coronary calcium test with a score of 0.

PMH: Hyperlipidemia, Fabry’s disease diagnosed in 2004, GERD.

Physical Examination: Alert, in moderate distress, neck supple, lungs clear, S1/S2 present and normal, no murmurs rubs or gurgles, no JVD or pedal edema.

Labs: White blood cell count 9.3 X 10^9/L, Hgb 12.9 g/dL, Platelet 363 X 10^9/L, troponin-0.013 => 0.175.

Diagnostcis: EKG done showed ST segment elevation in leads V2-V6 consistent with an early anterolateral infarct. Cardiac catheterization showed a 100% thrombotic occlusion of the mid LAD, treated with 3.5 x 23 mm Xience.

Hospital course:

- STEMI code was activated and the patient was sent to the Cardiac Catheterization Laboratory for a PCI with drug eluting stent placement and balloon angioplasty.
- The patient was started on dual antiplatelet therapy with aspirin and ticagrelor, high intensity statin and beta blocker.
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CARDIAC MANIFESTATIONS OF FABRY

- Left ventricular hypertrophy, aortic and mitral regurgitation, conduction defects, coronary artery disease, hypertension, and aortic root dilation.
- Among patients with Fabry’s disease, women are more likely to present with the cardiac variant of disease.
- 5.8% men and 3.7% women experience cardiovascular events at mean ages of 45 and 54 years, respectively.
- Examination of intramural coronary arteries on endomyocardial biopsy reveals luminal narrowing due to hypertrophy and proliferation of smooth muscle and endothelial cells with glycosphingolipid deposits.
- Heart failure was the most common first cardiovascular event.
- Endomyocardial biopsy useful when diagnosis is uncertain.
- Screening with enzymatic assay blood or leukocyte suggested for family members.
- Treatment: Enzyme replacement therapy in addition to standard therapies for heart disease.
- Observational studies suggest that ERT can reduce LVH and improve myocardial function.

CONCLUSION

- Timely commencement of enzyme replacement therapy has been demonstrated to contribute to regression of cardiac morbidities in FD, improvement in myocardial function and quality of life.
- Clinicians should have a high index of suspicion in the evaluation, timely detection and monitoring of cardiac pathologies in patients with FD.

REFERENCES