ATYPICAL PRESENTATION OF CARDIAC FABRY'S DISEASE

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CASE VIGNETTE

INTRODUCTION: Heart failure is typically the most commonly reported cardiovascular event in patients with Fabry's disease (FD) while ventricular arrhythmias and bradyarrhythmias are some of the causes of sudden cardiac death. Other notable cardiac findings are hypertrophic cardiomyopathy and valvular lesions with mitral insufficiency being the most common. We report the case of a patient with Fabry's disease who developed an ST elevation myocardial infarction (STEMI) with a 100% thrombotic occlusion of the mid left anterior descending (LAD) coronary artery.

CASE: A 68-year-old female with a history of FD diagnosed in 2004 managed with enzyme replacement therapy presented to the emergency department for evaluation of sudden onset heaviness/pressure-like discomfort in the center of her chest, 5/10 intensity, radiating down both arms with numbness and a tingling sensation, associated with shortness of breath, diaphoresis and lightheadedness. This was her first episode of such symptoms. She recently had a coronary calcium test with a score was 0. She reported a family history of FD in her father who also suffered from an unspecified heart problem. The patient reported a history of hyperlipidemia but denied hypertension, diabetes or chronic kidney disease. She was found to have ST segment elevations were noted in leads V2-V6 consistent with an early anterolateral infarct. Aspirin 324mg and 0.4mg sublingual nitroglycerin were administered and the patient was taken to the cardiac catheterization lab. Coronary angiography revealed a 100% thrombotic occlusion of the mid LAD. Balloon angioplasty was performed and a drug eluting 3.5mm x 23mm Xience stent was placed in the LAD.

DISCUSSION: Ceramide trihexidosis (Anderson-Fabry disease) is an X-linked lysosomal storage disease caused by deficient activity of the lysosomal enzyme alpha-galactosidase A. This enzymatic defect results in the accumulation of the glycosphingolipid (GL3) globotriaosylceramide in the lysosomes of vascular endothelium of several organs, smooth muscle cells and cardiac myocytes. The classic phenotype caused by the absent activity of α-gal A has an estimated prevalence of 1 in 40,000 males. Heterozygous females can be asymptomatic or present with cryptogenic stroke, renal failure or cardiac disease, with randomization of X-chromosomal inactivation accounting for the variable penetrance. Late-onset cardiac FD typically has a less severe phenotypic presentation compared to the classic. FD can be detected by newborn screening for α-gal A activity. It is plausible that the 100% LAD stenosis in our patient was due to the combination of hyperlipidemia and small vessel dx from endothelial GL3 accumulation.

CONCLUSION: Timely commencement of enzyme replacement therapy with an agent such as Fabrazyme has been demonstrated to contribute to regression of cardiac morbidities in FD and also improvement in myocardial function and quality of life. A high index of suspicion in the evaluation, timely detection and monitoring of cardiac pathologies in patients with FD is essential.