Introduction

- Mitochondrial Encephalopathy with Lactic Acidosis and Stroke-like Episodes (MELAS) is a maternally inherited mitochondrial syndrome typically diagnosed in childhood or early teenage years.
- The stroke-like episodes typically present in a relapsing-remitting manner with gradual neurological decline leading to dementia.

Case Presentation

- A 32 y/o man with a diagnosis of MELAS, confirmed by muscle biopsy 8 years prior, presented with a 2 month history of shortness of breath, dyspnea on exertion, and lower extremity edema.
- The patient was afebrile with a blood pressure of 108/90, heart rate of 103, respiratory rate of 20, and a room air oxygen saturation of 100%.
- Cardiac exam revealed a III/VI holosystolic murmur heard best at the apex and an S3.
- Lung exam was unremarkable.
- He had 3+ bilateral lower extremity pitting edema extending to his hips.
- Laboratory studies showed a WBC count of 5.7 K/uL with 71% neutrophils. His complete metabolic profile revealed HCO₃ 18 mmol/L, BUN 38 mg/dL, creatinine 1.67 mg/dL, bilirubin 3mg/dL, AST 92 U/L, ALP 213, U/L and ALT 100 U/L.
- BNP was greatly elevated at >5,000 pg/mL. Initial troponin was elevated at 0.14 ng/mL. Lactic acid was also elevated at 3.9 mmol/L.
- Urinalysis did not contain any protein or bacteria. Urine electrolytes revealed a FeNa of 6.7%; no urine eosinophils were present.
- EKG demonstrated normal sinus rhythm with evidence of left atrial enlargement, and chest x-ray revealed mildly increased pulmonary vasculature and an enlarged cardiac silhouette.

Hospital Course

- Diuresis with IV furosemide provided improvement in his respiratory status and edema.
- 2D echocardiogram showed 4 chamber enlargement, moderate to severe tricuspid regurgitation, an ejection fraction of < 20%, mild pulmonary hypertension, and multiple left ventricular apical thrombi.
- He was started on anticoagulation with fondaparinux and warfarin. After diuresis, he was started on carvedilol and an ace inhibitor was held until his creatinine stabilized.
- Hemoglobin A1C was found to be 6.5% and the patient underwent diabetes education.
- He was continued on his home MELAS treatment regimen during his hospital stay which included keppra, neurontin, co-enzyme Q, and L-carnitine.
- Upon discharge, he required home oxygen and home health was arranged.

Discussion

- MELAS is a rare mitochondrial disorder that not only affects the nervous system, but can also have cardiac, renal, and endocrine manifestations, as was seen in this patient with a dilated cardiomyopathy, renal insufficiency, and diabetes mellitus.
- A hypertrophic cardiomyopathy is more commonly associated with MELAS, but a dilated cardiomyopathy has also been described.

References