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654.MGUS, AMYLOIDOSIS AND OTHER NON-MYELOMA PLASMA CELL DYSCRASIAS: CLINICAL AND EPIDEMIOLOGICAL

GI Amyloid: More Than a Heavy Heart

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Amyloid transthyretin (ATTR) amyloidosis is caused by the accumulation of insoluble transthyretin (TTR) amyloid fibrils. These proteins can build up in various organ systems such as the musculoskeletal, gastrointestinal, and ocular system. However, there is usually a predilection for the heart and nervous system [1]. ATTR comprises approximately 16% of GI amyloid cases, with the small bowel being the most common area of involvement [2]. Symptoms typically include abdominal pain, nausea and vomiting, unintentional weight loss, diarrhea, and constipation. Given the rarity of the disease, we present a case of biopsy proven ATTR GI amyloidosis to raise awareness and, consequently, aid in early recognition. Our patient is a 48-year-old female with a medical history of substance use disorder who had multiple evaluations for complaints of chest discomfort and gastrointestinal symptoms including nausea, vomiting and diarrhea. Initial evaluation included an echocardiography which found severe left ventricular hypertrophy. Given her significant family history of myocardial infarction, a left cardiac catheterization was completed, however, this revealed normal coronary vasculature. Subsequently, the patient started medical therapy for heart failure with preserved ventricular function pending further testing. Despite medical management, the patient developed worsening symptoms of heart failure warranting repeat echocardiography which showed a significant reduction in ejection fraction. In addition to the patient's worsening cardiac symptoms, she continued to have refractory gastrointestinal symptoms and developed new epigastric pain. Therefore, the patient underwent upper endoscopy with discovery of abnormal peristalsis. After which, a gastric emptying study was done, resulting in a diagnosis of gastroparesis. With the constellation of findings i.e., the pattern of ventricular hypertrophy, lack of hypertensive and coronary artery disease, as well as gastroparesis without a known etiology, there was suspicion for systemic infiltrative disease, which led to further investigation. Gastric, small, and large bowel biopsies confirmed amyloid deposits. The patient also underwent a Tc99m-PYP scan which revealed evidence of cardiac amyloidosis; monoclonal protein analysis test was negative, and a cardiac biopsy showed a profile consistent with amyloid transthyretin type. The patient remained on pharmacologic therapy for gastroparesis and has been referred to cardiac transplantation services for further management.Our case highlights that amyloidosis, including GI amyloidosis, has the potential to go unrecognized due to non-specific signs and symptoms. Clinicians should therefore always have a high index of suspicion as GI involvement in systemic amyloidosis, regardless of protein subtype, is common. This high index of suspicion will allow for the appropriate pathologic examination to be pursued, and ultimately the appropriate management.

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