

FROM INTESTINAL ULTRASOUND TO A DIAGNOSIS OF HEREDITARY TRANSTHYRETIN AMYLOIDOSIS

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Poster

Colorectal

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A 67-year-old man with chronic diarrhea, orthostatic hypotension, malabsorption, and significant weight loss was diagnosed with hereditary transthyretin amyloidosis after extensive investigation revealed gastroparesis, hypertrophic cardiomyopathy, and genetic confirmation of a transthyretin gene mutation.

KEY POINTS

- The patient presented with chronic diarrhea from 2019, orthostatic hypotension, malabsorption, and weight loss of around 20 kgs.
- Entero-MRI and videocapsule showed gastroparesis associated with rapid small bowel transit; echocardiography and cardiac cine-MRI revealed hypertrophic cardiomyopathy with accumulating disease.
- Abdominal fat aspirate enabled the diagnosis of amyloidosis, and genetic testing confirmed a heterozygous mutation in the transthyretin gene.
- Hereditary transthyretin amyloidosis comprises 16% of amyloidosis subtypes found in the GI tract and causes peripheral neuropathy with autonomic features, cardiomyopathy, or mixed phenotype.
- The authors note that hereditary transthyretin amyloidosis is difficult to diagnose in early stages and is frequently misclassified as other digestive diseases, emphasizing the importance of early diagnosis given recently approved therapies.

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