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Summary

UNDER AN EXCEPTIONALLY RARE PRESSURE: PORTAL HYPERTENSION IN MAHVASH DISEASE

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Pancreas

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A 54-year-old man with massive pancreatic enlargement was diagnosed with Mahvash disease, a rare genetic disorder caused by GCGR gene variants, presenting with a well-differentiated pancreatic neuroendocrine tumor and subsequent portal hypertension.

KEY POINTS

- Genetic evaluation revealed likely pathogenic variants c.1176+1_1176+7del p.? and c.361T>C p.(Cys121Arg) in the GCGR gene (heterozygous), confirming Mahvash disease, with only about 15 confirmed cases reported to date.
- Biopsy confirmed a well-differentiated (G1) pancreatic neuroendocrine tumor with amyloid stroma; PET/CT with 68Ga-peptides showed focal uptake in the pancreatic tail (SUV max 47.8) and increased uptake throughout the pancreas.
- Treatment was initiated with octreotide and later transitioned to lanreotide.
- Two years later, the patient developed portal hypertension with liver nodules characteristic of hypervascular metastases and large esophageal varices with red spots, which were treated with prophylactic elastic band ligations.
- The case is comprehensively documented through multimodal imaging (CT, PET, MRI, endoscopic ultrasound) and histology, illustrating a rare genetic cause of pancreatic neuroendocrine neoplasm and portal hypertension.

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