Neurofibromatosis Type 1 With Ampullary Somatostatinoma, Small Bowel GISTs, and Extra-Adrenal Composite Pheochromocytoma-Ganglioneuroma

Patrick Hickey DO  
*Lehigh Valley Health Network, Patrick.Hickey@lvhn.org*

Rohit Sharma MD  
*Lehigh Valley Health Network, Rohit.Sharma@lvhn.org*

Shanth A. Goonewardene MD  
*Lehigh Valley Health Network, Shant_A.Goonewardene@lvh.com*

Shashin Shah MD  
*Lehigh Valley Health Network, Shashin.Shah@lvhn.org*

Hiral N. Shah MD  
*Lehigh Valley Health Network, hiral_n.shah@lvhn.org*

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Neurofibromatosis Type 1 With Ampullary Somatostatinoma, Small Bowel GISTs, and Extra-Adrenal Composite Pheochromocytoma-Ganglioneuroma

Patrick Hickey, DO, Rohit Sharma, MD, Shanth Goonewardene, MD, Shashin Shah, MD and Hiral Shah, MD
Lehigh Valley Health Network, Allentown, PA

Background
- Neurofibromatosis type 1 (NF1) is an autosomal dominant hereditary tumor syndrome
- Increased risk of GI tumors including neurofibromas, GISTs, and periampullary carcinoids is reported
- Periampullary somatostatinomas as well as true neurogenic tumors including extra-adrenal pheochromocytomas (paragangliomas) and ganglioneuromas have also been reported
- This case study reviews a patient with NF1 and multiple intra-abdominal neoplasms

Initial Case Description
- A 32 year old male presented with six months of jaundice, dark urine, and weight loss
- CT: dilated intrahepatic ducts, and elevated bilirubin and ALP
- ERCP with biopsy of the ampulla: invasive malignancy with neuroendocrine differentiation
- Pancreatoduodenectomy showed low-grade somatostatinoma of the ampulla and an incidental duodenal GIST

Case Continuation
- CT scan at year two found an enlarging upper abdominal mass concerning for nodal metastasis
- Ex-lap removal found a paraganglioma in association with ganglioneuroma
- Given the pathology, the diagnosis of NF1 was confirmed with mutation analysis showing a truncating mutation in NF1
- His exam showed multiple café au lait spots on the trunk and upper extremities, multiple neurofibromas, and axillary freckling
- Years later he had a progressive right adrenal nodule and elevated plasma/urine metanephrines
- Resection of a paracaval functional paraganglioma and partial right adrenalectomy with postoperative XRT to the right adrenal bed was performed
- One year later, CT and OctreoScan showed a tumor corresponding with nodularity in the jejunal limb and pancreaticobiliary anastomosis
- Surgery confirmed proximal and distal jejunal GISTs
- Systemic octreotide treatment is under consideration

Discussion
- Alterations in the NF1 gene (tumor suppressor) encodes for the protein neurofibromin
- Unlike sporadic GISTs, NF1 associated GISTs do not usually harbor c-KIT or PDGFRα mutations
- Pheochromocytomas reportedly affect 0.1-6% of NF1 patients. 3/14 reported cases of composite pheochromocytoma-ganglioneuroma were associated with NF1
- The combination of pheochromocytoma and GIST has been described in 14 NF1 cases
- Periampullary tumors in NF1 patients are most commonly carcinoid, but most do not develop somatostatinoma syndrome
- To our knowledge this is the first case of a patient with NF1 diagnosed with ampullary somatostatinoma, small bowel GISTs, and extra-adrenal composite pheochromocytoma-ganglioneuroma occurring together

References:

Figure 1: Paraganglioma Intermediate Power. Figure 2: Paraganglioma High Power.