



Neurofibromatosis-Related Biliary Obstruction: Is There A Role In Screening For Biliary Malignancy?

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LEARNING OBJECTIVES

1. Review cancer screening guidelines for NF1 patients
2. Present and discuss the need for additional screening

CASE PRESENTATION

A 63-year-old female with NF-1 presents with a 5-month history of abdominal pain, nausea, and a 40-lb weight loss was found to have ileocecal thickening and intra/extrahepatic biliary ductal dilation on imaging.

EVALUATION

- CEA & CA19-9 normal
- EGD showed multiple large polyps in the second portion of the duodenum
- ERCP was notable for a periampullary mass causing biliary obstruction and a mid-CBD stricture that was biopsied and stented across (Figure 1)

COURSE/RESULTS

- Pathology revealed ganglioneuroma and was negative for malignancy.
- Ileocecal thickening was found to be colon adenocarcinoma
- The patient underwent right hemicolectomy and chemotherapy.
- Subsequently, she had recurrent episodes of cholangitis related to periampullary obstruction and numerous biliary stents.

COURSE/RESULTS

- EGD/ERCP at 1 year showed villous appearance of the ampulla with surrounding congestion. An irregular biliary stricture was again appreciated with pathology revealing ganglioneuroma

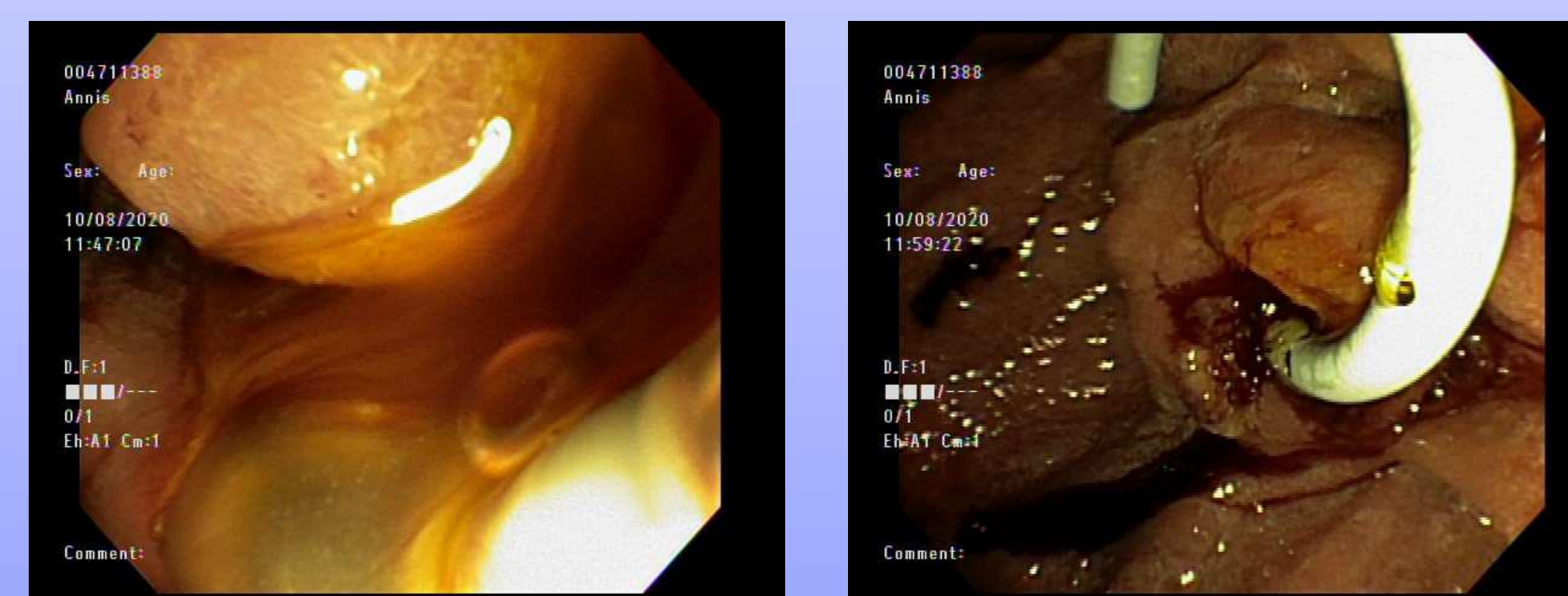


Figure 1. Demonstration of periampullary mass and mid-CBD stricture

REFERENCES

Hirbe AC, Gutmann DH (2014) Neurofibromatosis type 1: a multidisciplinary approach to care. *Lancet Neurol* 13(8):834–843

Agaimy A, Vassos N, Croner RS (2012) Gastrointestinal manifestations of neurofibromatosis type 1 (Recklinghausen's disease): clinicopathological spectrum with pathogenetic considerations. *Int J Clin Exp Pathol* 5(9):852–862

Levy AD, Patel N, Dow N, Abbott RM, Miettinen M, Sobin LH (2005) Abdominal neoplasms in patients with Neurofibromatosis type 1: radiologic-pathologic correlation. *Radiographics* 25(2):455–480

DISCUSSION

NF1 has known associations with numerous malignancies

- Breast, bone, thyroid, liver, lung, colon, ovarian, esophageal, NHL, rhabdomyosarcoma, leukemia, CNS tumors

Other than early initiation of breast cancer screening, there are no other specific cancer screening guidelines for NF patients compared to the general population, despite the known high-prevalence of concomitant tumors in these patients

Up to 25% of NF1 patients can have GI tract involvement

- But only 5% are symptomatic

NF-1 patients with periampullary or biliary tract masses have the potential for malignant transformation.

- There are no screening guidelines to facilitate early detection of tumor progression in this population

Patients with NF-like findings should also be considered to have other potential underlying genetic mutations

- Constitutional mis-match repair deficiency increases the risk of malignant transformation by ~80%

CONCLUSION

Our patient had numerous ERCPs over one year

- Although last ERCP had a villous appearance of her periampullary mass, biopsy remained negative for malignancy
- She will undergo repeat sampling and stent removal, where biopsy will be repeated

We suggest the need for biliary imaging/sampling to screen for malignant transformation in similar patients