International Journal of Allied Medical Sciences and Clinical Research (IJAMSCR)

ISSN: 2347-6567

IJAMSCR |Volume 3 | Issue 3 | July-Sep- 2015
www.ijamscr.com

Case Report *Medical research*

Neuroendocrine tumour of periampullary region presenting as obstructive jaundice in a patient of neurofibromatosis: a case report

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ABSTRACT

Patients of neurofibromatosis sometimes develop neuroendocrine tumour [NET]. These NETs are usually found in duodenum or periampullary region .We report a case of a patient with neurofibromatosis, who presented with obstructive jaundice. Upper gastrointestinal (UGI) endoscopy showed a periampullary growth. An endoscopic biopsy was taken which was not conclusive of any neoplastic growth. A pylorus preserving pancreaticoduodenectomy was performed. The specimen was sent for histopathology. The tumor was later on diagnosed as well differentiated NET, low grade.

KEY WORDS: Neurofibromatosis, Neuroendocrine tumor, Obstructive jaundice, Pancreaticoduodenectomy

INTRODUCTION

Neurofibromatosis-1 (NF-1), also known as von Recklinghausen's disease, is an autosomal dominant, multisystem disorder that occurs in 1 in 2,500–3,000 live births [1,2]. This hereditary disorder is characterized by pigmentary features (caféau-lait macules, skinfold freckling), neurofibroma, orthopedic features (scoliosis, dysplasia of a long bone), and ophthalmologic features (Lisch nodules, optic glioma) [1-3]. The NF-1 gene is located on chromosome 17q11.2, and encodes the protein neurofibromin [4,5]. Neurofibromin is a tumor suppressor expressed in many cells, so NF-1 gene mutation leads to diminished level of neurofibromin which subsequently causes uncontrolled cell proliferation and development of benign and malignant tumors, including neuroendocrine tumors (NETs) [1,3]. Neuroendocrine tumours (NETs) are slow-growing malignancies derived from the neuroendocrine system. Sixty per cent of NETs arise in the gastrointestinal tract [6], less than 1% of these occur in the ampulla of Vater and 25% of these cases can be associated with neurofibromatosis. Ampullary NETs are usually non-functioning. Most are well-differentiated neoplasms with low malignant potential [7,8]. The most frequent target organ of NETs in patients with NF-1 is the duodenum and peri-ampullary region. We report a unique case of a patient with generalized neurofibromatosis presenting with obstructive jaundice due to periampullary NET.

CASE PRESENTATION

A 42 year old patient with NF1 presented with jaundice for 3 months with complain of mild epigastric pain and passing clay colored stool. There was no history of hypoglycemia or peptic ulcer disease. On clinical examination, there were miltiple cutaneous neurofibromas scattered over whole body with a large plexiform neurofibroma over sacral area. The patient´s BP was 128/74 mmhg and heart rate was 78/min. Serum total bilirubin was 2.18mg/dl with conjugated bilirubin

1.24mg/dl, suggestive of obstructive jaundice. Ultrasonography of abdomen showed dilated CBD and IHBR with mildly dilated pancreatic duct, tiny peripancreatic lymph nodes with mild hepatosplenomegaly. Subsequently, an endoscopic retrograde cholangio pancreaticograhy [ERCP] was performed which showed a periampullary growth. A biopsy was taken from growth endosopically and sent for HPE and stent was inserted. The report was not conclusive of any neoplastic growth.CECT abdomen was suggestive of subtle periampullary pathology with multiple lymphadenopathy with distended gallbladder filled with sludge. Later on, a pylorus preserving pancreaticoduodenectomy and cholecystectomy was performed with hepaticojejunostomy, pancreaticojejunostomy and gastrojejunostomy. Intraoperatively, a nodular growth was palpabe in periampullary region. The specimen (fig.1) was sent for HPE (fig.2). The patient made excellent postoperative recovery. The biopsy report was conclusive of well differentiated neuroendocrine tumor, low grade with tumor size: 2.0 cm and 1.1 cm in greatest dimension, located in periampullary area and gastroduodenal junction invading muscularis propria. Pancreatic, common bile duct, proximal gastric and distal duodenal margins were negative for tumor. Surrounding lymph nodes were negative for tumor as well. The tumor was staged pT2N0Mx according to AJCC/TNM stage.

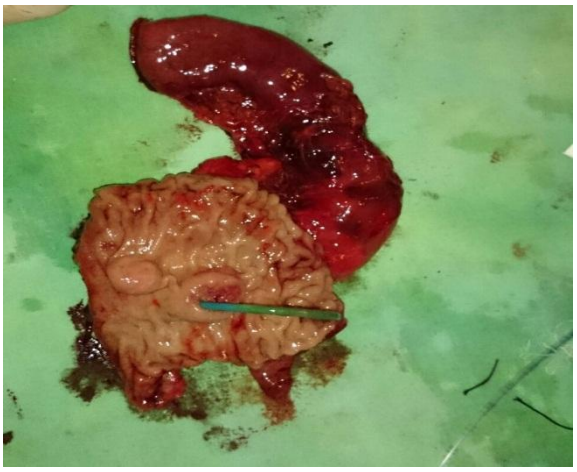

Fig 1: Resected specimen showing periampullary growth with stent .

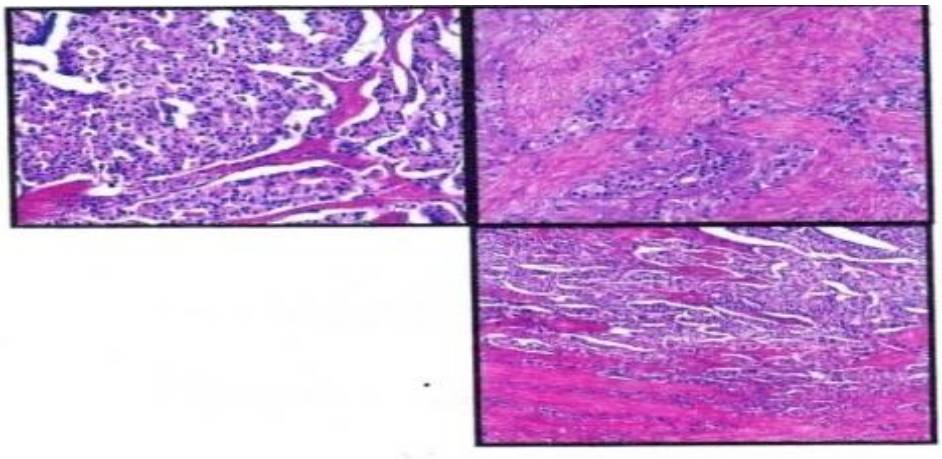

Fig 2: Microscopic picture of resected specimen (low power view).

DISCUSSION

NF1 is one of the most common neurogenetic diseases affecting adults and children. It is a complex progressive disease affecting multiple cell types and multiple systems of the body. Diagnostic criteria for NF1 highlight these diverse manifestations and include pigmentary lesions, neurofibromas, optic pathway gliomas and bony dysplasias [3]. NF1 is transmitted as an autosomal dominant disorder caused by mutations in the NF1 gene on chromosome 17. The NF1 gene encodes a protein: neurofibromin, which functions as tumour suppressor by inactivating the RAS molecule and preventing its mitogenic signaling, resulting in reduced cellular proliferation. Mutations at the NF1 gene result in diminished levels of neurofibromin that may predispose to subsequent development of a variety of tumours [5]. Ampullary NETs have been described in association with NF1 [8] and may present as obstructive jaundice or as acute pancreatitis. The NET in the present case had a low mitotic and proliferative index (Ki67), normal plasma chromogranin, small size and no concomitant liver metastases. Other additional favorable features were male sex, absence of bone metastases or ectopic hormone production.

CONCLUSION

Periampullary neuroendocrine tumor is rare in patients of neurofibromatosis. It has better prognosis than adenocarcinomas.Pancreaticoduodenal resection is procedure of choice for periampullary NETs.

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How to cite this article: Mokarram Ali. MD, Kajla, Prabhu Dayal .R.K, Pradeep Bhalerao, Rakesh Khedar, Neuroendocrine tumour of periampullary region presenting as obstructive jaundice in a patient of neurofibromatosis: a case report. Int J of Allied Med Sci and Clin Res 2015; 3(3):285-287.