

## Case Report

# Ampullary Ganglioneuroma: an unusual feature of Neurofibromatosis Type 1 - A Case Report

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### Abstract

The case of a 65 year old man is presented. He had a history of dyspepsia and was diagnosed as neurofibromatosis clinically. Upper G.I. endoscopy showed thickened folds in duodenum. A polyp was seen at the junction of D1 and D2 adjacent to the ampulla. Duodenal biopsy results were suggestive of ganglioneuroma.

### Introduction

Neurofibromatosis is an autosomal dominant disorder. The main signs are café-au-lait spots on the skin and cutaneous neurofibromas. The disorder is subdivided into type 1 or von Recklinghausen's disease, type 2 or central neurofibromatosis and other types.<sup>1</sup> Type 1 neurofibromatosis can involve any organ system and involvement of the gastrointestinal tract has been well documented.<sup>2</sup>

### Case Report

A 65 year old male presented with the complaints of

hiccups, epigastric pain and dyspepsia for the last 8 years. On physical examination, multiple nodular lesions were noted over the limbs, neck, chest and forehead (Figure 1). Rest of the examination was unremarkable. Clinical diagnosis of neurofibromatosis was made and upper G. I. Endoscopy performed to find out the cause of his symptoms. Endoscopy showed thickening of folds in the duodenum. A one cm broad based polyp was seen at the junction of D1 and D2 adjacent to the ampulla. Ampulla was also bulky with raspberry appearance (Figure 2).

Figure 2. Endoscopic appearance of bulky polypoid ampulla.

Biopsies were taken from polyp and ampulla. Patient was advised for barium follow through and colonoscopy to exclude intestinal polyps. Barium follow through did not show any additional polyps. Lower G.I. endoscopy up to terminal ileum was also normal.

The duodenal biopsy showed spindle cell proliferation in the lamina propria with the fibrillary background. Scattered foci of mature ganglion cells were also identified. Features were suggestive of ganglioneuroma (Figure 3).

Figure 3. High power photomicrograph showing a cluster of mature ganglion cells embedded in a spindle cell background.H&Ex400.

### Discussion

Neurofibromatosis type 1, also called von Recklinghausen's disease is the most common form of neurofibromatosis and one of the most common autosomal dominant disorders in man.<sup>3</sup> It arises from the mutation of NF1 gene located on chromosome 17q.<sup>4</sup> About 25% of individuals affected with neurofibromatosis type1 exhibit multiple intestinal polypoid neurofibromas or less commonly ganglioneuromas. The small bowel is affected most often, followed by the stomach and finally the colon.<sup>5</sup>

Intestinal ganglioneuromas are an overgrowth of nerve tissue in the mucosa, submucosa, or muscle layers of the gastrointestinal tract. The overgrowth may be polypoid or cause diffuse thickening of the bowel wall. Intestinal

ganglioneuromatosis has been observed in neurofibromatosis type 1.

The actual incidence of these tumors, which tend to be multiple in the gastrointestinal tract, is uncertain because the diagnosis is suspected only in those patients who have symptoms. A significant number of cases with neurofibromatosis type1 never manifest gastrointestinal bleeding, obstruction or abdominal pain, the three most common presenting symptoms with this group of lesions. Our patient presented with dyspeptic symptoms and was found to have ampullary lesion incidentally on endoscopy. The biopsy of ampulla showed features of ganglioneuroma. He did not have any signs or laboratory tests suggestive of biliary obstruction, so ERCP was not done.

It can be concluded that the gastrointestinal dysfunction, in the setup of von Recklinghausens disease mandates complete endoscopic and radiologic evaluation of the digestive tract to find out or rule out neurofibroma or gastrointestinal stomal tumor. Surgical resection is the cornerstone of treatment in patients who present with obstruction or gastrointestinal bleeding.

### References

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