THE CONCURRENT OCCURRENCE OF MULTIPLE POLYPOSIS COLI WITH HIRSCHSPRUNG DISEASE IN A SINGLE PATIENT

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ABSTRACT

We report the concurrent occurrence of Hirschsprung disease and multiple polyposis coli on barium enema confirmed with histopathology in a 7 year old child who presented to the Civil hospital Karachi with history of chronic constipation. Hirschsprung disease is the most common cause of colonic obstruction in a pediatric age population. The polyposis syndromes are rare disorders in which more than 100 gastrointestinal polyps are present throughout the GI tract. We present the rare case of concurrent occurrence of these two diseases of different etiology and pathogenesis in a single patient.

Key words: Hirschsprung disease, Barium enema, and polyposis coli.

Introduction

Hirschsprung disease is a developmental disorder characterized by absence of ganglia in the distal colon, resulting in a functional obstruction. Most cases of Hirschsprung disease are diagnosed in the newborn period.1 It is thought to either occur from a failure of neuroblasts in neural crest cells to migrate into bowel segments or degeneration of already migrated neuroblasts. It affects cells both in the myenteric and submucosal plexuses.2 On the other hand, polyposis syndrome is characterized by the appearance of multiple polyps in the gastrointestinal tract. Polyps are abnormal growths arising from a mucous membrane.3 Although intestinal polyposis syndromes are relatively rare, awareness of the existing health risks is important for patients and their families affected by these disorders.

Case Summary

A 7 year-old Asian male is presented to Civil hospital with complaints of chronic constipation on and off since birth relieved only with laxatives and kleen enema. There was no history of weight loss, bleeding per rectum or pus/mucoid discharge. He has two elder sisters and 1 younger brother; none has similar complains. The family history revealed no such issues existing in the family. The physical examination was unremarkable except for mild pallor. On admission, laboratory investigations showed hemoglobin 10 g/dl, white blood cell count 5.33 × 10⁹ /L, platelet count 118 × 10⁹ /L and mean corpuscular volume 77.8fl. He was referred to the radiology department for barium enema which apart from changes of Hirschsprung disease (Fig. 1) i.e. narrowing at the recto sigmoid junction, collapsed rectum, inversion of the recto sigmoid index, and grossly distended sigmoid colon and descending colon; demonstrated multiple innumerable barium coated smooth well defined and broad based as well as pedunculated intraluminal filling defects (polyps) diffusely distributed throughout the colon (Fig. 2), largest at the anti-mesenteric border of transverse colon measures 34 x 28 mm. Colonoscopy with biopsy from the largest polyp shows hamartomatous tissue containing mucous glands and connective tissue with no evidence of dysplasia or malignant cells.
Discussion

Intestinal polyposis syndromes are broadly classified into hereditary and non-hereditary types. The hereditary types include hereditary non polyposis colorectal cancer, familial adenomatous polyposis syndrome (FAPs), Gardner syndrome, Turcot syndrome, Cowden syndrome, Peutz-Jeghers syndrome and non-hereditary include hyperplastic polyposis syndrome, Cronkhite-Canada syndrome. In juvenile polyposis syndrome, approximately 25% cases are familial and 75% are non-hereditary in origin.

Juvenile polyposis syndrome is a rare syndrome characterized by hundreds of distinct juvenile hamartomatous polyps in the gastrointestinal tract containing fluid/mucous and an increased risk of colorectal cancer. The term “juvenile” refers to the type of polyp rather than to the age of onset of polyps.\(^4\)_5

In 2010 Whittle DO et al\(^6\) reported the case of Juvenile polyposis syndrome in a 21-year-old male with a two-year history of intermittent rectal bleeding. Colonoscopy revealed multiple pedunculated cherry red polyps mainly in the left colon. Histology confirmed juvenile polyps.

Sodhi KS et al\(^7\) in 2005 documented a case of a 5-year-old boy with intermittent rectal bleeding. Barium enema X-ray showed multiple rectal and colonic polyps. Histological evaluation revealed multiple juvenile polyps without adenomatous change or dysplasia.

Ikeda Set al\(^8\) in 2009 reported the first case of Hirschsprung's disease associated with familial adenomatous polyposis (FAP) in a 14-year-old Japanese female who had multiple polyps in her large intestine, stomach, and duodenum. Five members of her family had been diagnosed as having FAP.

The aim of our case is to document the rare coexistent occurrence of multiple polyposis coli and Hirschsprung disease. These are two entirely different diseases, each having specific origin and pathogenesis. Each deserves special attention, as each disease can have different sequelae. For example polyposis coli can lead to colorectal carcinoma after a latent period.\(^6\) The affected individuals also need genetic counseling and surgical intervention is the
only means of cure for Hirschsprung disease (HS). Our patient was planned for surgery of HS, and afterwards he was kept on surveillance, and interval colonoscopies with follow up visits were recommended.

References


