CASE REPORT

Gardner's syndrome, a rare combination in surgical practice

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SUMMARY

A 45-year-old man presented to the emergency ward with features of intestinal obstruction of 2 days duration. On admission, there was abdominal distension and multiple sessile polyps found on digital rectal examination. In addition, a soft tissue swelling near the elbow and a bony swelling over scalp were noted. Abdominal radiography revealed gaseous distension of the small and large bowel, and ultrasound revealed diffuse, gas-filled bowel with sluggish peristalsis. The obstruction failed to resolve with conservative measures and at emergency laparotomy an irregular hard recto-sigmoid junction mass was identified. A defunctioning transverse loop colostomy was undertaken and the abdomen closed. During recovery, a colonoscopy was performed and a malignant appearing lesion was identified 15 cm proximal to the anal verge. Further per-stomal colonoscopy revealed multiple sessile polyps from the ileo-caecal valve to the descending colon. The cutaneous and abdominal findings were consistent with a rare acute presentation of Gardner's syndrome.

BACKGROUND

Devic and Bussy first described what is now known as Gardner's syndrome (GS) in 1912 as a triad of intestinal polyposis, various soft tissue tumours such as fibromas, lipomas, neurofibromas and epidermoid cysts, and multiple osteomas particularly involving the skull. The syndrome is named after Eldon J Gardner a geneticist, who first described it in 1951.¹ GS is an autosomal dominant form of polyposis characterised by the presence of multiple polyps in the colon together with extra-colonic tumours. Extra-colonic manifestations include osteomas of the skull, thyroid cancer, epidermoid cysts, fibromas and sebaceous cysts, dental abnormalities and periampullary carcinomas as well as the occurrence of desmoid tumours in approximately 15% of affected individuals. The incidence of the syndrome is 1 in 14 000 with an equal sex distribution.² Inheritance is autosomal dominant and it arises because of mutations in the adenomatous polyposis coli gene (APC) on chromosome 5q21.³ Polyps may also arise in the stomach, duodenum, spleen, kidneys, liver, mesentery and small bowel. The number of polyps increases with age, and hundreds to thousands of polyps can develop in the colon. GS is also known as familial adenomatous polyposis (FAP) and may later manifest as an aggressive fibromatosis (desmoid tumours) of the retroperitoneum, which are much more difficult to treat.⁴

Figure 1 Photograph showing osteoma over the forehead.

CASE PRESENTATION

A 45-year-old man presented to the emergency ward of our institution with features of intestinal obstruction of 2 days duration. On admission the pulse, blood pressure and other observations were normal; however, there was a marked distension of the abdomen with visible peristalsis, but palpation revealed the abdomen to be soft and non-tender. The liver was not enlarged and spleen impalpable. Bowel sounds on admission were absent and digital rectal examination demonstrated multiple sessile polyps with associated ballooning and tenting of the rectal mucosa. General examination revealed a hard bony swelling over the scalp and the back of the pinna was suggestive of an osteoma (figure 1) and a further soft tissue swelling near the wrist was consistent with an epidermoid cyst (figure 2). A family history was taken and it revealed that the

Figure 2 Photograph showing epidermoid cyst over the right forearm.

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patient's father died of a gastrointestinal malignancy but further details were not known.

INVESTIGATIONS

On emergency admission, the full blood count identified a haemoglobin level of 9 g/dL; bleeding time and clotting time, serum electrolyte and blood urea nitrogen were normal. Plain abdominal radiograph revealed gaseous distension of the small and large bowel (figure 3). Ultrasonography revealed diffuse, gas-filled bowel with sluggish peristalsis, a minimal fluid collection in the pelvis and a sonographically normal liver.

On further evaluation after initial defunctioning surgery, colonoscopy was performed per rectum identifying a malignant appearing lesion at 15 cm above the anal verge. Perstomal colonoscopy revealed multiple sessile polyps extending from the ileo-caecal valve to the descending colon with a large tumour mass at the recto-sigmoid junction showing surface mucosal necrosis (figure 4) biopsy of which revealed adenocarcinoma. The serum carcino embryonic antigen level was 14.62 ng/mL.

DIFFERENTIAL DIAGNOSIS

The gastrointestinal findings raise the possibility of a number of differential diagnoses which are associated with colonic polyposis including:

- ▶ FAP
- ▶ Peutz-Jeghers syndrome
- ▶ Turcot syndrome
- ▶ Juvenile polyposis syndrome
- ▶ Cowden disease

TREATMENT

In this case transverse loop colostomy was performed as an emergency to relieve the impending obstruction. On re-exploration after confirmation of diagnosis formal resection could not be safely undertaken because of extensive loco-regional and nodal spread and extensive polyposis of the gastrointestinal tract distal to the ileo-caecal valve.

Figure 3 Plain abdominal radiograph showing distended bowel loops of the large and small gut.

Figure 4 Perstomal colonoscopy showing multiple colonic polyps and recto-sigmoid growth.

OUTCOME AND FOLLOW-UP

After re-exploration wound healing was delayed, the patient was discharged from the hospital 3 weeks after the re-exploration but sadly died of progressive disease complicated by community-acquired pneumonia 9 weeks after discharge from hospital.

DISCUSSION

This combination of multiple polyps in colon together with epidermoid cyst, osteoma over scalp signifies towards GS, which is a rare combination in surgical practice. In this case a preoperative CT scanning of the abdomen and pelvis to evaluate operability prior to second surgery could not be undertaken because of the patient's limited finances.

About half of the patients with GS develop adenomas by 15 years of age and 95% by age 35 years.⁵ Generally, cancers start to develop a decade after the appearance of the polyps and if the colon is left intact, the majority of patients with FAP will develop colorectal cancer (CRC) by the age of 40–50 years. However, it should be emphasised that, although uncommon, CRC can develop both in children and in older adults, where restorative proctocolectomy with ileal pouch should be considered if the syndrome is diagnosed at an early stage. After restorative surgery upper gastrointestinal (GI) tract screening by endoscopy for duodenal polyposis and treatment of desmoids by surgery and cryoablation are well established.

Osteomas, the most benign major feature of GS, are fortunately the first sign to be noticed by the patient or by the parents of an affected child and should precipitate further investigation.⁶ It is reported that approximately 25% of patients with GS have no family history.^{7–9} Delayed or incorrect diagnosis of these patients is common particularly if there is no obvious family history or previously detected colorectal polyps. In addition, those patients who do demonstrate polyposis should have their duodenum, thyroid, teeth, skulls and eyes examined to rule out associated pathology.

Screening recommendations include flexible proctosigmoidoscopy at the age of 10–12 years; repeat every 1–2 years until the age of 35; after the age of 35 years repeat every 3 years. Upper GI endoscopy should be performed every 1–3 year starting from when polyps are first identified.¹⁰ It is reported that non-

steroidal anti-inflammatory drugs (NSAID) can prevent the progression of colorectal adenomas in patients with FAP and hereditary non-polyposis colorectal cancer (HNPCC) and therefore NSAID use may prevent the polyp in FAP (including GS) and HNPCC.^{11–16}

Learning points

- ▶ A possible combination of sign and symptoms should always be considered in day-to-day surgical practice. Those who have a positive family history of colonic malignancy should undergo screening with periodic colonoscopy, upper GI endoscopy.
- ▶ Suspicious cases should undergo genetic screening for early detection and prophylactic treatment. Prophylactic proctocolectomy should be considered in high-risk groups.
- ▶ A nationwide documentation of these cases should be made, so as to spread awareness among the health professionals as well as patients.

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