Management of infantile polyposis syndrome

G J Stiff, A Alwafi, H Jenkins, J Lari

Abstract
A boy with generalised infantile polyposis syndrome is reported to highlight the difficulties in management. Despite attempts to reduce polyp mass by regular endoscopic polypectomy, daily transfusions of blood products, and a trial of the non-steroidal anti-inflammatory agent sulindac, his condition gradually deteriorated and he died of septicaemia.

(Arch Dis Child 1995; 73: 253–254)

Keywords: infantile polyposis syndrome.

The term ‘juvenile polyp’ was coined by Horriheno and his colleagues to describe the characteristic histological appearance of polypoid hamartomas arising from the lamina propria of the colorectum within the paediatric age group.1 McColl and his coworkers are generally credited with the first description of multiple juvenile polyps and used the term juvenile polyposis syndrome to distinguish these patients from those with a solitary juvenile polyp.2 There are three forms of this rare condition in which polyps are limited to the colon (juvenile polyposis coli), distributed throughout the entire gastrointestinal tract (generalised juvenile polyposis), or present in infancy either as colonic or diffuse disease (infantile polyposis syndrome, IPS).

We report a case of IPS affecting the whole of the gastrointestinal tract and describe the management difficulties associated with this condition. We also report the use of the non-steroidal anti-inflammatory agent sulindac that was used in an attempt to reduce the polyp mass.

Case report
A boy of 7 months of age presented with rectal bleeding, abdominal pain, and bile stained vomiting. There was no past history of abdominal symptoms and no family history of gastrointestinal disease. On examination there was a palpable abdominal mass in the left lower quadrant and a full blood count showed iron deficiency anaemia with a haemoglobin concentration of 86 g/l. A barium enema gave normal results but an ultrasound scan strongly suggested the diagnosis of intussusception.

A laparotomy was performed which confirmed jejunojejunal intussusception with a large 4 cm polyp leading the intussusception. In addition a large number of polyps were palpable in the rest of the small bowel as well as in the colon. Numerous polyps were removed through the enterotomy and histology showed the typical appearances of juvenile polyps. A diagnosis of the IPS was established.

Postoperatively he was noted to be oedematous and liver function tests revealed an albumin concentration of 18 g/l. He was given several infusions of salt poor albumin but his serum proteins remained low and therefore total parenteral nutrition was initiated.

Other clinical features of note on examination included generalised hypotonia, macrocephaly with a head circumference well above the 95% centile, clubbing of his fingers and toes, and increasingly obvious alopecia. These have been previously noted in IPS. A colonoscopy performed a month post-operatively showed hundreds of polyps of varying size that were distributed from the caecum to the rectum. In excess of 20 polyps were snared and sent for histology. Despite regular twice monthly colonoscopic removal and diathermy of hundreds of polyps he continued to suffer bloody diarrhoea and mucus per rectum, weight loss, and polyps prolapsing from the rectum. Despite total parenteral nutrition and regular transfusions of blood, albumin, fresh frozen plasma, and platelets it proved impossible to correct the severe hypoalbuminaemia, hypogammaglobulinaemia, and anaemia.

At the age of 15 months upper gastrointestinal barium studies and endoscopy confirmed the presence of a large number of polyps in the stomach and small bowel that varied in size from a few millimetres to several centimetres. He underwent a further colonoscopy at this time that showed hundreds of new medium and large polyps despite having a polypectomy three weeks previously which had cleared in excess of 40 polyps.

In view of the limited surgical options he was started on the non-steroidal anti-inflammatory drug sulindac (Clnoril, MSD) in the hope that it would help reduce the polyp mass. Literature reports had documented its success in the management of familial adenomatous polyposis and there was a single report of its use in IPS. The patient was treated for six months at a dose of 50 mg daily. Further colonoscopies showed no regression but a continuing increase in the numbers of polyps. The various treatment options, including small bowel transplantation, had been discussed at length on several occasions with the child’s parents but in view of his poor general state this was not felt to be appropriate. He suffered frequent infections and septicaemic episodes that responded to intravenous antibiotics and also required surgical excision of several polyps that caused troublesome rectal prolapse. However, despite intensive treatment with daily infusions of whole blood, platelets, and albumin he subsequently developed frequent haematemeses and eventually died at the age of 18 months from overwhelming septicaemia.

Discussion
IPS is an inherited condition that is not sex linked and may be associated with the presence of congenital abnormalities such as Meckel’s diverticulum, malrotation, pulmonary arteriovenous malformations, hypertrophic osteoarthropathy, and cardiac anomalies. There may also be macrocephaly, hypotonia, clubbing, and alopecia as seen in this case. Individuals with this condition usually present before 2 years of age with anaemia due to blood loss caused by rectal bleeding in association with malabsorption and hypoproteinanaemia due to persistent diarrhoea. Alternatively they may present with a complication of polyposis such as intussusception, prolapse, or intestinal obstruction and it is the poor nutritional status that is responsible for the high morbidity and mortality seen in this group of patients.

Management of IPS, in particular the generalised form, should be conservative and follow a format similar to that of generalised juvenile polyposis syndrome. The effects of blood and protein-rich fluid loss in an infant are greater than in older children, however, and individuals such as our child may require daily transfusions of blood, platelets, and albumin to keep up with gastrointestinal losses. A poor nutritional state and loss of plasma proteins (including immunoglobulins) may predispose the child to the development of central catheter sepsis that may prove difficult to treat. There is little to offer in the way of surgical options unless the child develops obstruction, intussusception, or an irreducible prolapse. One option that may be available is intestinal transplantation, although patients with infantile polyposis are generally in a poor nutritional state and are not ideal candidates for such major surgery, particularly when the whole of the intestine is affected as in our child.

One possible medical treatment that has been proposed is the drug sulindac, a non-steroidal anti-inflammatory drug which has shown to be effective in reducing the size and number of colonic and rectal adenomas in familial adenomatous polyposis and in Gardner’s syndrome. Sulindac has been shown to inhibit cell proliferation of adenomas and it is hypothesised that this may be through reducing the synthesis of prostaglandin E2, which is found in high concentrations in such tumours. There is a single case report of the successful use of sulindac in juvenile polyposis syndrome which described its administration to a 2 year old boy with generalised disease. The authors reported a reduction in both size and number of polyps when the drug was administered daily over a period of three months. We treated our patient with a dose of 50 mg daily (5 mg/kg) but we did not see any reduction in size or number of the polyps. Recent correspondence with the authors of the first case report revealed that despite the apparent early success, sulindac failed to control the polyps in the long term. Thus the role of sulindac in IPS remains uncertain and the outlook for these children remains very poor.

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Arch Dis Child 1995 73: 253-254
doi: 10.1136/adc.73.3.253

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