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PEUTZ-JEGHERS SYNDROME: CASE REPORT
G.O. Igun (FWACS), Y.Y. Ameh, (MBBS) and K.U. Awani, (MBBS), Department of Surgery, University Teaching Hospital, Jos, Nigeria.

Request for reprints to: Dr. G.O. Igun, Department of Surgery, University Teaching Hospital, PMB 2076, Jos, Nigeria

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SUMMARY

A case of Peutz-Jeghers syndrome (PJS) in an African girl is presented to document the clinical presentation, management and follow up of this condition. The patient who presented with black buccal mucosal hyperpigmentation and clinical features of jejuno-jejunal intussusception was successfully managed by operative reduction of the intussusception and polypectomy. She is being followed up for evidence of malignant transformation in associated intestinal polyps and development of extra-intestinal malignancies at other sites. The management of PJS in light of recent trends is discussed, especially with reference to suggested protocols for screening and surveillance of sites at supposed risk of tumour development.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is a rare, autosomal dominant disease entity whose clinical presentation is well known. Peutz and later Jeghers(1) described the syndrome characterised by small intestinal hamartomatous polyps and hyperpigmented macules of the lips and other areas. The overt melanotic macules of PJS often appear during the first one or two years of life and may increase in number and distribution with fading with age of all lesions with the exception of buccal lesions in some cases. It is, however, the intestinal polyps that first draw attention to the existence of this syndrome because of associated complications like ulceration, bleeding and partial intestinal obstruction due to intussusception(2). There has been a vexing debate about the risk of gastrointestinal malignancies in patients with PJS and also isolated case reports of its association with extra intestinal cancers. All these facts call for a review into the cause, clinical features, natural history, management and surveillance programmes that form the subject of this case report.

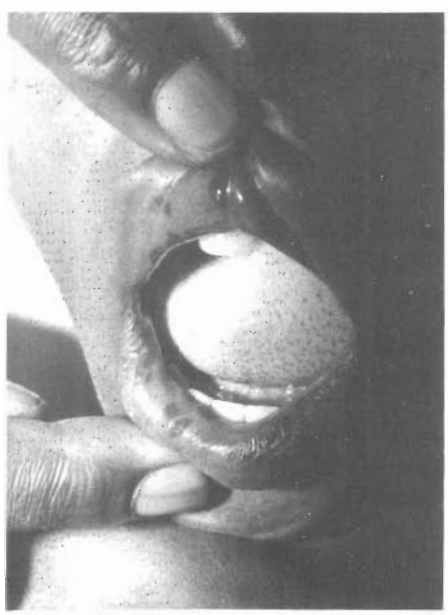
CASE REPORT

M.N., a 12-year old female student presented at the outpatient clinic with severe generalised colicky abdominal pain of five months' duration. The pain was associated with occasional vomiting of recently ingested food. She was anorexic, had lost some weight and also complained of melaena stools. She sought medical help at the beginning of her ailment from a rural hospital where appendicectomy was performed but her symptoms persisted. There was no history of similar illness in her family.

On examination, she was found to be ill-looking, mildly-pale, not dehydrated or jaundiced. She had a systemic blood pressures of 120/100mmHg and a pulse rate of 100/minute. She had black mucosal hyperpigmentation of her lower lips with dark-brown pigments in both palms and soles of her feet (Figure 1). Abdominal examination revealed a flat abdomen with a post-appendicectomy scar and visible peristalsis from left to right. There was a sausage shape tender mass that measured 10cm by 6cm situated in the left periumbilical region of her abdomen. Bowel sounds were hyperactive and digital anorectal examination revealed an empty rectum with melaena stools on the gloved finger.

Figure 1

Baccal macosal hyper-pigmentation in PJS



Her urea and electrolyte results showed no abnormality, packed cell volume was 32%. She was subjected to elective exploratory laparotomy through a lower midline incision one week after presentation. The findings at exploration of the abdomen were a viable, reducible, jejuno-jejunal intussusception with a pedunculated polyp measuring 4 by 4cm. There were also other pedunculated polyps of about the same size that were palpated 40cm from the duodeno-jejunal

flexure and three other that were situated in the sigmoid colon. The offending polyp was exercised from the base following traction on the head, after manual reduction of the intussusception (Figure 2). The abdomen was closed employing the mass closure technique.

Figure 2

Polypectomy specimen in PJS

Postoperatively, the patient was placed on prophylactic intravenous ampicillin 250mg six hourly, gentamicin 40mg eight hourly and metronidazole 250mg eight hourly for three days. She made an uneventful recovery, sutures were removed on the tenth postoperative day and she was discharged for follow up care in the outpatient clinic. Histopathology reports of the excised polyp revealed a hamartomatous, smooth muscle polyp with normal brush border epithelium, without cellular atypia.

DISCUSSION

The most recent concept of PJS is that it is one of the familial hamartomatous polyposis of the gastrointestinal tract, the other being juvenile polyposis. It is a rare syndrome which is encountered about one tenth as often as adenomatous polyposis like Gardener's syndrome and Turcots syndrome(3). Gene mapping studies are still underway to identify the chromosomal location or locus of the responsible DNA defect. Males/female ratio in PJS is about 1:1. Black or dark-brown pigmented macules occur as a distinctive feature of the disease in more than 95% of affected sons(4). They are perioral in distribution in (94%), occur on the palms (74%), buccal mucosa (66%)(3), soles, digits, peri-anal area and rarely in the intestinal mucosa. Hamartomatous polyps occurs anywhere in the gastro-intestinal tract apart from the mouth. They are most commonly found in the small intestine (64-94%) but also occur in the stomach and colon. Polyps are found in (88-100%) of patients with PJS. Symptoms directly related to the polyps occur in the second and third decade from infraction, ulceration, bleeding (14%), intestinal obstruction and jejuno-jejunal intussusception (43%) as in the patient we have presented.

Insight into the natural history of these intestinal polyps indicates that adenomatous change and malignant transformation occur in two to thirteen per cent of patients with PJS(4). Bailey's(2) estimated risks of gastrointestinal malignancy of 24% was probably an over-estimation due to misinterpretation of pseudoinvasion in histopathology specimen or reported fortuitous cancer association. The weight of present day evidence also favours mortality figures approaching 40% with these cancers by 40 years of age(5). Several extra-intestinal tumours, both benign and malignant have also been reported to have an association with PJS from isolated case reports based on small series of patients. In women, these include bilateral breast carcinomas, cervical adenocarcinomas and ovarian tumours, especially benign sex cord turmours. In males, there is an association with Sertoli cell tumours. Pancreatic adenocarcinomas also occurs in both genders in patients with PJS.

All these call for strict surveillance in the management and subsequent follow up of patients with PJS including the use of such ancillary investigations as yearly haemoglobin estimations and abdominal ultrasonography. Biannual small bowel barium follow-through studies and "top plus tail" endoscopy are also needed including mammography in female patients until they are fifty years of age. Three yearly cervical smears are also required in female patients. In addition, apart from the exploratory laparotomy especially indicated for intussusception, preoperative enteroscopy and polyectomy is required for diagnosis and treatment of residual polyps of the gastrointestinal tract especially if they are more than 1.5cm in diameter(5). The cost-effectiveness of the various management options can only be described in relative terms. Screening programmes aimed at early case detection of intestinal and extraintestinal cancers have the advantage of prompting early surgical intervention if such cases are discovered. The prognosis for patients such treated is far better than for those managed for advanced cancers. All these gains will reflect in better survival figures for patients with PJS and appreciable financial and manpower gains for those countries that embark on such surveillance programmes. These programmes will also decrease unnecessary laparotomies for large polyps that are fraught with such complications like bleeding and intestinal obstruction Subsequent emergency re-laparatomies for obstructions due to adhesions from previous surgery will also be reduced. This proposed management protocol for patients with PJS can therefore to be seen as a highly effective cost-effective, long term measure aimed at the overall treatment of the phenotypic manifestations of the rare genetic inherited syndrome of the disease.

REFERENCES

1. Jeghers, H., Mc. Kusick, U.A. and Katz, K.H. Generalized intestinal polyposis and melanin sports of the oral mucosa, lips and digits. A syndrome of diagnostic significance N. *Engl. J. Med.* 1949; **241**:1031-6.
2. Bailey, D. Polyposis of the gastrointestinal tract: The Peutz-Jeghers syndrome. *Brit. Med. J.* 1957; **2:** 483-9.
3. Desai, D.C., Neale, K.F. and Talbot, I.C. *et al.* Juvenile Polyposis. *Brit. J. Surg* 1995; **82**:14-17.
4. Yamada, T. Textbook of gastro enterology. In: Randall, R.W., eds. Peutz-Jerghers Syndrome. Philadelphia: J.B. Lippincott Co. 1991; 1684-85.
5. Spidelman, A.D., Arese, P. and Phillips, R.K.S. Polyposis: The Peuty-Jegher syndrome. *Brit. J. Surg.* 1995; **82**: 1311-14.