Unusual manifestations of the Peutz-Jeghers syndrome

Intestinal polyposis with mucocutaneous pigmentation was first described by the Dutch physician Peutz in 1921.¹ In 1949 Jeghers² assembled 22 definite and nine possible cases from published reports. We present a patient with severe anaemia secondary to gastrointestinal haemorrhage. The site of bleeding was multiple jejunal hamartomas of the type classically found in the Peutz-Jeghers syndrome. The features of interest are, firstly, the lack of mucocutaneous pigmentation, and, secondly, the restriction of all identifiable hamartomas to the jejunum. A search of published work has not shown a case with these hamartomatous malformations without in addition mucocutaneous pigmentation.

Case report

A 19-year-old English man presented with iron-deficiency anaemia. Five years previously he had been found to be anaemic (Hb 5.7 g); the results of tests for faecal occult bloods had been positive but barium studies had failed to disclose a source of gastrointestinal bleeding. He had responded to treatment with oral iron and had remained well for four years, when he again developed gross iron-deficiency anaemia, while contrast radiological examination of the gastrointestinal tract again failed to demonstrate any abnormality. Six months later radiochromium studies indicated that he was losing substantial amounts of blood into the gut, a loss of about 570 ml of blood being estimated in one week. He repeatedly denied any gastrointestinal symptoms, complaining only of lethargy and weakness. He was clinically anaemic but there was no mucocutaneous pigmentation. Repeat investigations showed no abnormality apart from the iron-deficiency anaemia, and coeliac and superior mesenteric arteriograms failed to demonstrate any abnormality. The findings on gastroscopy and sigmoidoscopy were also within normal limits. There was no family history of similar complaints.

Laparotomy disclosed several large polyps in the proximal jejunum (figure), which extended through the proximal five feet of jejunum, which was resected. Histological examination of the polyps showed hamartomatous lesions compatible with the Peutz-Jeghers's syndrome characterised by smooth-muscle components not found in adenomatous polyps.³ The patient made an uneventful postoperative recovery and has remained well.

A hamartomatous polyp.

Discussion

We report this case because of the absence of mucocutaneous pigmentation, the lack of a family history, and the prolonged and severe bleeding. In a review of 75 cases of Peutz-Jeghers's syndrome Bartholomew⁴ found pigmentation on the lips of all but one and the buccal mucosa of all but six. Melanin pigmentation of the skin and mucous membrane characteristically appear in infancy and early childhood. The lips are affected almost universally, and the buccal mucosa in 83%; 30% have pigment on other facial orifices, fingers

or toes, or perianal area. With advancing years the pigmented freckles tend to fade. It has been said that when carefully searched for mucosal pigmentation is invariably found in patients with this type of intestinal polyposis, whereas cutaneous pigmentation may be missing.

The most probable genetic explanation for the syndrome is a single pleiotropic gene responsible for both characteristics. The likelihood of the polyposis and the pigmentation being caused by two closely linked genes is remotely possible but no instance of crossing over within a family has been recorded. The single gene probably does not have complete penetrance, as pedigrees with skipped generations have been published and there are far too many isolated cases for all of them to be due to fresh mutation.

Of 59 patients with polyps studied by Rivers *et al*,⁵ 14 had minor gastrointestinal tract bleeding, haemorrhage being thought to complicate episodes of intussusception by the polyps.

It would appear that the Peutz-Jeghers syndrome may have several forms frustes as well as the complete clinical picture, and the description of an incomplete spectrum occurring in isolation may be of interest genetically.

We wish to thank Dr Irvine Delamore for referring the patient and Dr R B McConnell for his help and advice.

¹ Peutz, J L A, *Nederlandsch maandschrift voor geneeskunde*, 1921, **10**, 134.

² Jeghers, H, McKusick, V A, and Katz, K H, *New England Journal of Medicine*, 1949, **241**, 933, 1031.

³ Morson, B S, *Diseases of the Colon and Rectum*, 1962, **5**, 337.

⁴ Bartholomew, L G, Dahline, D C, and Waugh, J M, *Gastroenterology*, 1957, **32**, 434.

⁵ River, L, Silverstein, J, and Tope, J W, *International Abstracts of Surgery*, 1956, **162**, 1.

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Neurological presentation of psittacosis during a small outbreak in Leicestershire

In 1975, 177 cases of chlamydia B infection (psittacosis) were reported to the Public Health Laboratory Service (personal communication), but more must go undetected in view of the mild illness often produced. The usual presentation is of an influenza-like illness with prominent respiratory symptoms, but dominant cerebral symptoms appear less common. We wish to emphasise this aspect in a patient seen recently during an outbreak of psittacosis in Leicestershire.

Case report

In June 1975 a 49-year-old woman was admitted to the Leicester Royal Infirmary. Two weeks previously she had developed an irritating dry cough and a week later she developed headache, fever, anorexia, right-sided pleuritic chest pain, a worsening mental state, and facial asymmetry. Her speech became rambling, her concentration and memory were poor, and she was hallucinating, apathetic, and oblivious of her illness. Her husband owned a pet shop, and three of his birds had recently died, including a sick red lory (species of psittacine).

On examination she was drowsy, disorientated, and confused. She was pyrexial (40°C), tachypnoeic, and her pulse was 120 per minute. There were crepitations over a small area of the right lower chest posteriorly, a right supranuclear facial palsy, and extensor plantar responses. Investigations showed normal blood count, cerebrospinal fluid, and electrocardiogram, an erythrocyte sedimentation rate of 90 mm/h, and right lower lobar consolidation on chest radiography.

Because of her contact with ill psittacine birds tetracycline 500 mg six-hourly was given. She made a dramatic response, her temperature falling to normal within 24 hours and chest radiograph clearing after nine days, but her mental state took four weeks to return to normal.

The complement fixation (CF) tests for influenza A and B, mycoplasma, and Q fever were negative. Serum CF titre for chlamydia B was less than 1/8

on admission, 1/8 two weeks later, and 1/256 seven weeks after the onset of her illness. Her husband's titre was 1/64, and he had had a severe influenza-like illness soon after his shop opened in December 1974.

Discussion

Our patient's neurological presentation was consistent with a diagnosis of encephalitis, and this was reported in only two of 156 cases in 1972.¹ Clearly reliance cannot be placed on an early negative CF test for psittacosis to exclude the disease in an unusual presentation, and in addition the administration of tetracycline may delay the appearance of a rising titre for several weeks.

From September 1974 to July 1975 10 cases of psittacosis were documented at the Public Health Laboratory in Leicester. Together with two other cases² all had some connection with a particular pet shop dealer in Leicestershire, who himself developed a respiratory illness in 1974 after importing some Bolivian parrots. One patient merely helped in moving an aviary and his birds to a new house, and, of the others, our patient and her husband stocked their shop from this dealer, one bought directly from him, and two were wives of bird enthusiasts who also bought from the dealer.

Since the ban on importation of psittacines was lifted³ unknown numbers of birds infected with *Chlamydia psittaci* have entered Britain. The practice by dealers of giving tetracycline reduces the high mortality in recently imported infected birds, but partially treated birds may survive to excrete organisms for months. Repeated warnings about this zoonosis have been given,⁴ and we were surprised to find that many pet shop owners and private aviarists were still unaware of the importance of the disease. A quarantine for imported parrots and other birds was reintroduced on 1 March 1976.⁵

We are grateful to Professor R Kilpatrick for permission to report a patient under his care.

¹ *British Medical Journal*, 1973, **3**, 704.

² Walker, C W M, *State Veterinary Journal*, 1975, **30**, 206.

³ Parrots and Miscellaneous Birds (Prohibition of Importation Revocation) Order, 1966.

⁴ Keymer, I S, *Lancet*, 1973, **2**, 1437.

⁵ Importation of Captive Birds Order, 1976.

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Syphilis presenting as hair loss

The diagnosis of syphilis may be considerably delayed when the manifestations are unusual, single, or modified by inadequate treatment¹: and even when in retrospect they are typical.² The two cases reported here were referred to a dermatology department solely on account of hair loss. They were shown to have syphilis, the treatment of which resulted in regrowth of hair.

Case reports

Case 1—A 24-year-old woman had noted progressive loss of scalp hair for three months. At the beginning of this time she had had a sore throat, cough, and headache, but at no stage did she have a rash. Eight weeks before beginning to lose hair she had had sexual intercourse with a stranger while on holiday in Greece. On initial examination she had slightly uneven thinning of scalp hair, but no abnormalities of the hairshafts or scalp (figure). The results of skin, mucous membranes, body hair, and general examination were normal. The results of investigations were Venereal Diseases Research Laboratory test: positive 1:4; fluorescent treponemal antibody absorption and treponemal haemagglutination: positive; telogen count: top of scalp

Case 1. Uneven hair loss (left) and regrowth two months after treatment (right).

35%, side 61% (samples about 80 hairs, normal less than 30%). Histopathology of scalp showed sparse chronic inflammatory cell infiltrate in the upper dermis. She was treated with procaine penicillin 600 000 U daily for 10 days, and her hair began to regrow six weeks from the onset of treatment (figure). The telogen counts returned to normal.

Case 2—A 25-year-old homosexual man presented because of thinning of scalp hair for four months. Two months before this he had a slightly itchy rash consisting of small macules and papules mainly on the trunk, which had cleared after three weeks. At no time had he felt ill. He had no relevant medical history, and a routine serological check for syphilis had given a negative result 18 months earlier. There was widespread thinning of scalp hair, although, as in case 1, the decreased density was not uniform. Hair elsewhere was unaffected, and the results of clinical examination, including proctoscopy, were otherwise normal. The results of investigations were VDRL: positive 1:32; FTA-ABS: strongly positive; TPHA positive. In the cerebrospinal fluid both the VDRL and TPHA were positive. Telogen count: top 43%, side 14%. Histopathology of the skin showed a sparse lymphocytic infiltrate around the superficial aspects of several hair follicles. Because of the positive serological reactions in the spinal fluid he was treated with procaine penicillin, 600 000 U for 21 days. His hair began to regrow four weeks after starting treatment.

Discussion

Both these patients had their alopecia reversed by effective treatment of the syphilis. Both had self-limited conditions which may have been part of their illness: upper respiratory symptoms in case 1 and a rash in case 2. Nevertheless, in neither case did these episodes suggest syphilis. These cases differ in that case 1 was at the secondary stage, and case 2 had asymptomatic neurosyphilis, although the clinical and histological features of their alopecia were similar.

Could the alopecia be unrelated to syphilis? The hair loss was less dramatic than the "moth eaten" appearance illustrated in many textbooks, but nevertheless patchy. Histologically the changes were not typical of syphilitic alopecia,³ although consistent with both this and alopecia areata.⁴ Nevertheless, alopecia areata seems unlikely in view of the response to treatment. Despite the high telogen count, the patchy hair loss and presence of inflammation are against a diagnosis of telogen effluvium. Hence it would seem prudent to consider syphilis in patients with non-scarring alopecia which cannot otherwise be readily explained.

I am grateful to Dr S C Gold for permission to report case 1, Dr A G Knight for permission to report case 2, and Dr R C Catterall, Director of James Pringle House, Middlesex Hospital, where both cases were referred for treatment.

¹ *British Medical Journal*, 1972, **3**, 66.

² *British Medical Journal*, 1975, **2**, 460.

³ Wile, U J, and Belofe, G H, *Archives of Dermatology and Syphilology*, 1926, **13**, 495.

⁴ Van Scott, E J, *Annals of the New York Academy of Science*, 1959, **83**, 480.

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