Diagnosis may be difficult even with resected specimens of bowel (Schmidt *et al.* 1968) and serial sections may be required to find the granulomas.

If the diagnosis is known, medical treatment is indicated. Should the symptoms thereafter be severe enough to warrant surgery, then the procedure must be carefully chosen.

Lockhart-Mummery (1972) advocated abdominoperineal excision of the rectum and terminal colostomy when disease was localized to the sigmoid colon and rectum. Where the colon was extensively involved, but the rectum was normal, ileorectal anastomosis was considered unsafe; a better procedure was total proctocolectomy and ileostomy.

It is concluded that Crohn's disease must be suspected when a slightly atypical colonic disease presents in an older person, or when a straightforward bowel resection is followed by major complications. Mucosal biopsies are unreliable at present in making a diagnosis, but will become more important in the future due to the increasing use of fibreoptic colonoscopes.

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Mr I Burn (*Charing Cross Hospital*) queried the statement that diarrhœa was commonly a symptom of colonic diverticular disease. The presence of this symptom generally implied other coincidental pathology or malfunction.

Dr J S Stewart (*West Middlesex Hospital*) asked about the studies of intestinal transit time in diverticular disease being carried out in Reading. What, for instance, were the effects of adding bran to the diet?

Mr A J Brodribb (*Battle Hospital, Reading*) said he was studying a group of patients with uncomplicated diverticular disease in Reading. A significant proportion had a rapid colonic transit time and complained of frequent loose motions. On adding bran to the diet colonic transit time slowed and motions became firmer in this group of patients.

The following cases were also presented:

Tuberculosis Associated witn Multiple Myeloma
Dr F M Andrews

Sjögrens's Syndrome with Rheumatoid Arthritis, Neuropathy and Chronic Active Hepatitis
Dr F P Bresnihan

Meeting 7 June 1974

Cases

Ehlers-Danlos Syndrome Presenting with Torsion of Stomach

M P Linnemann BA MB (for V W Johnson MB MRCP)
(New Cross Hospital, Wolverhampton WV10 0QP)

B R, woman aged 68

History: Presented in 1969 aged 63, with pain in left hypochondrium, vomiting of four days' duration and features compatible with Ehlers-Danlos syndrome (EDS); not previously diagnosed. Two similar episodes occurred within two years and on each occasion she responded to conservative treatment with intravenous fluids and nasogastric aspiration. She had had a kyphoscoliosis for as long as she could remember and wounds had always taken a long time to heal. Dislocated right elbow at age 16. Over the years 3 thyroid adenomata excised followed by treatment with thyroxine. Osteoarthrosis affecting particularly the left knee first occurred at 45 and at 55 she complained of dyspnœa on exertion, and a high left diaphragm which did not move on screening was noted. At 60, she sustained multiple fractures of the left elbow and the radial head was excised, following which there was excessive soft tissue swelling.

On examination: Skin soft and hyperextensible. Joints of hands hypermobile. Marked kyphosco-

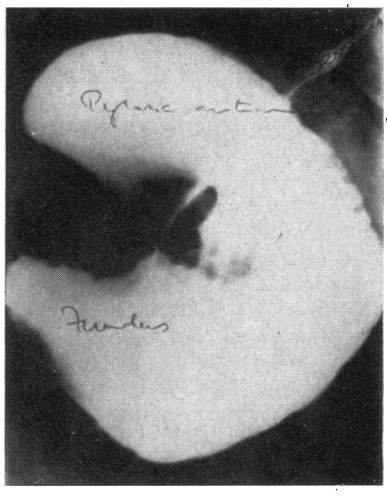

Fig 1 *Barium meal showing organo-axial torsion of the stomach*

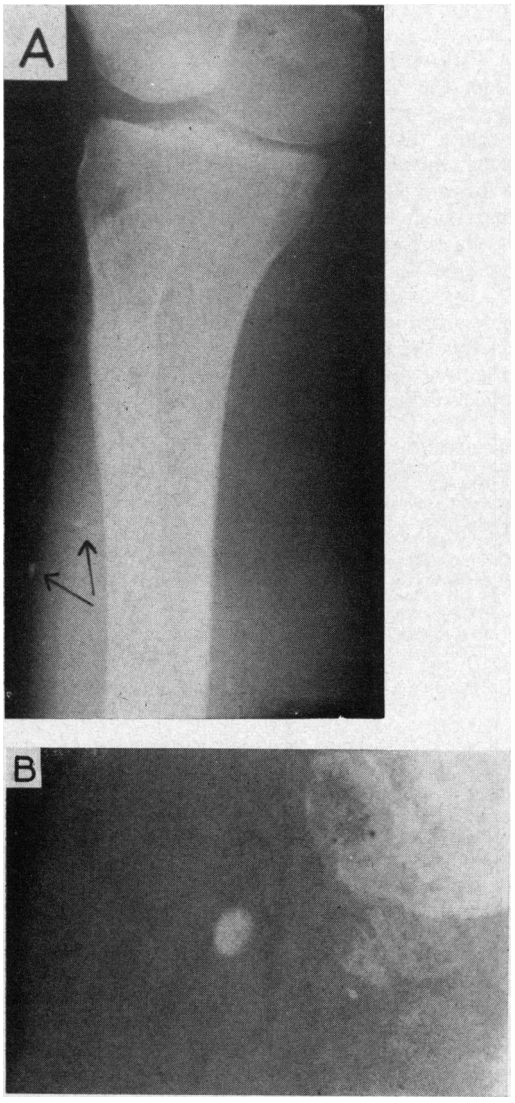

Fig 2 *Calcified subcutaneous spheroids*

liosis with osteoarthrosis of left knee, pes planus, bilateral varicose veins and an absence of striæ gravidarum. Incisional scars in neck; post-traumatic scars over both knees. Expansion of left hemithorax limited, with hyper-resonant percussion note at left base; breath sounds were absent on that side except at the apex. Abdomen normal.

Investigations: Chest X-rays showed eventration of left diaphragm; torsion of stomach suspected. Barium meal, performed on recovery from first attack, showed torsion of stomach about longitudinal axis with no delay in gastric emptying (Fig 1). Following recovery from the two subsequent attacks, barium meals showed the same appearances with minor delay in gastric emptying, despite the fact that the symptoms had settled. Barium enema demonstrated a few sigmoid diverticula.

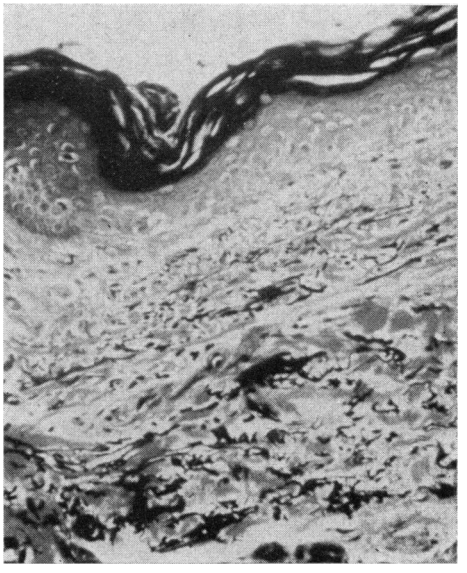

Fig 3 *Skin histology to show irregularity and loss of usual tramline appearance of collagen fibres. The elastin is stained black and clumped. van Gieson.* × *225*

The diagnosis of EDS was supported by the radiological demonstration of subcutaneous spheroids (Fig 2A, B) which are considered to be calcified ischæmic fat lobules and pathognomonic of this condition but should be differentiated by their shape and position from cysticerci and phleboliths (Beighton 1970). The spheroids were not palpable in our patient. Skin biopsy was performed although both histology and electron microscopy are notoriously unhelpful in establishing the diagnosis of this syndrome which is made on clinical grounds. The biopsy was not conclusive but van Gieson and trichrome stains showed that the collagen fibres were disorganized and lacked the normal regular tramline appearance; the elastin was unusual in that it was clumped (Fig 3).

We later elicited a strong family history of stigmata of EDS. The family tree (Fig 4) illustrated an autosomal dominant mode of inheritance. The patient's mother and maternal grandmother were both reported to have hypermobility of the joints of their hands and hyperextensible skin which bruised readily and healed slowly. One of the patient's brothers whose skin and joints were similarly affected died (aged 60) of Hodgkin's disease. Another brother who died (aged 65) was also affected and his daughter has

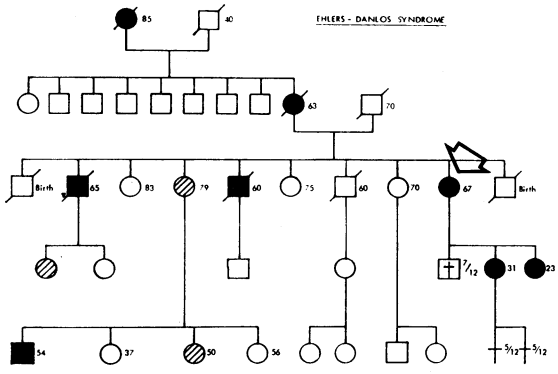

Fig 4 *Symbols used in the pedigree: the open circles represent normal females; the open squares normal males; the black circles are affected females; the black squares affected males; hatched symbols show presumed affected individuals; crosses indicate abortions and stillbirths; and diagonal lines are deceased persons*

congenital blindness. An elder sister of our patient (aged 79) had precocious osteoarthrosis and has a hiatus hernia while her daughter (aged 50) was admitted to hospital last year with a carotid-cavernous fistula and her son (aged 54) suffers from recurrent dislocation of the right elbow. Our patient was one of 10 children of whom the first and tenth siblings were both stillborn and possibly premature. Her son was stillborn at 7 months and both her daughters have hypermobility of the joints and hyperextensible skin. One daughter has pigmented papyraceous scars (including a skin graft) on both knees and as yet she has not achieved a successful pregnancy, having had two abortions, each at five months. This reflects the previously observed hazards of abortion and stillbirth in EDS reported by Bauer (1952), Graf (1965) and Beighton (1970) and suggests either that the fetus was severely affected or that there was connective tissue disorder of the chorionic membranes or cervical os.

Comment

Despite the eponym suggested by Parkes Weber in 1936 it seems undisputed that the first complete description of the syndrome was given by Tschernogubow in Moscow in 1891 when he presented a 17-year-old patient (sex not specified) with soft elastic skin and marked scarring on the face, knees and elbows, who also had dislocations of the left elbow and left hip which followed falls during childhood. He concluded that the connective tissue laxity involved the whole body.

From our patient's family tree it appears that the syndrome 'breeds true', the skin being principally affected, followed by hypermobility of the joints (especially the hands) while two members of the family have gastrointestinal features.

Previously recorded associations between EDS and the gastrointestinal tract include hiatus hernia, peptic ulceration, perforation, hæmorrhage, diverticulosis (Beighton *et al.* 1969) and megaduodenum and malabsorption syndrome (Hines & Davis 1973) but gastric torsion has not previously been reported in this condition.

Gastric torsion may be predictable in other members of the family by performing serial chest X-rays to detect eventration of the diaphragm. It is worth mentioning that surgery is best avoided in this syndrome because of the tissue fragility. Finally, it is clear that the syndrome has had no effect on life expectancy in this family.

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Dr V W Johnson (*New Cross Hospital*) said that surgical intervention in these patients had frequently been accompanied by tearing out of sutures because of tissue fragility and they should be treated conservatively whenever possible.

Dr B I Hoffbrand (*Whittington Hospital*) said there was an association between hiatus hernia and kyphoscoliosis. Edmunds (1957, *Quarterly Journal of Medicine* **26,** 445) had found an incidence of up to 60% of spinal deformities in a series of cases of rolling hiatus hernia. In a personal series of 11 consecutive cases of intrathoracic stomach seen on a general medical unit no fewer than 6 had had an associated kyphoscoliosis. He suggested that in the present case the diaphragmatic defect was related to the kyphoscoliosis which was in its turn due to the ligamentous abnormalities of the Ehlers-Danlos syndrome. He asked whether the affected relative with a hiatus hernia had a spinal abnormality.

Dr Linnemann replied that this relative did not have a spinal deformity.

Mr P S Phatak (*Whittington Hospital*) asked if it was known where the transverse colon lay in this woman. In cases of eventration of the diaphragm the transverse colon filled the extra space and rotated the stomach with it, producing the volvulus; in fact Tanner (1968, *American Journal of Surgery* **115,** 505) described an operation for the disconnexion of the colon from the stomach.

Dr Linnemann replied that the barium enema showed that the splenic flexure was within the space created by the eventration.

(meeting to be continued)