Gardner's syndrome and steatocystoma multiplex

Two unusual genetically determined conditions occurring in same patient

Summary. A 43-year-old man is described who had Gardner's syndrome and steatocystoma multiplex. These two unusual genetically determined conditions were associated because he had inherited the Gardner's syndrome from his father and the steatocystoma multiplex from his mother.

Gardner’s syndrome (Gardner, 1951) and steatocystoma multiplex (Klausner, 1917) are two uncommon conditions which are both inherited as autosomal dominant traits and they have not previously been described in the same patient. The following report of a family (Fig. 1) shows how this occurred in the proband, because he inherited Gardner's syndrome from his father and steatocystoma multiplex from his mother.

Report of a family

Ascertainment. The proband was examined at St Mark's Hospital where he had been diagnosed as having polyposis coli. A clinico-genetic study on families with polyposis coli and Gardner's syndrome was being carried out by one of us (B.L.) to determine which type of skin cyst occurred in Gardner's syndrome and whether familial polyposis and Gardner's syndrome were the same or different diseases. Members of 15 other families with Gardner's syndrome were seen and 60 affected individuals were examined and none of these showed steatocystoma multiplex. After the proband was seen, his father's hospital notes were reviewed and his mother and his children were visited in their homes. His sister had lived in the United States of America since her marriage, but full clinical details were obtained from her physician in that country.

Clinical features

III.1 R.C. Male aged 43 years. The proband had noticed cysts on his skin since the age of 8 years, and six had been removed since the age of 17 years. These were typical epidermoid cysts and were situated on the scalp (2), forehead, left thigh, right shin, and left ankle.

At the age of 22 years he first noticed small skin coloured nodules on the front of his chest and abdomen and these have not changed since that time (Fig. 2). Clinical examination showed 10 small (0.5–0.8 cm), bluish nodules attached to the skin, but freely moveable over the deeper tissues. They did not have a punctum (Fig. 3). There were none on his back, in his axillae, and groins or on the scrotum.

He first presented at St Mark's Hospital at the age of 30 years with a 10-year history of diarrhoea and a 10-month history of rectal bleeding. A diagnosis of polyposis coli was made on sigmoidoscopic examination and this was confirmed after the biopsy of a rectal polyp. A barium enema showed that the polyps extended through-
III.2 L.M. Female aged 38 years. From the age of 20 years to 26 years she had four children born by caesarian section. At the age of 23 years she developed a desmoid tumour in her abdominal wall scar which was removed. Four years later she developed rectal bleeding and was found to have polyposis coli. An abdomino-perineal resection was carried out removing the rectum, sigmoid, and left side of the colon. No cysts or osteomas were noted at that time and recent letters to her family say that she has no skin cysts.

II.1 N.C. Male aged 59 years. He was first seen at another hospital at the age of 33 years with a history of rectal bleeding for four years. A sigmoidoscopic examination showed a large carcinoma of the rectum, and an abdomino-perineal resection of the rectum was performed. There was no mention of colonic and rectal polyps in the clinical notes or in the histology report. Eighteen years later, at the age of 51 years, he was referred to St Mark's Hospital with bleeding from his colostomy. A barium enema showed polyps of the ascending colon and a total colectomy with ileostomy was performed.

He died suddenly at the age of 59 years with a sub-arachnoid haemorrhage. There was no necropsy. His wife said that he developed a lump on his left forearm at the age of 35 years, which was removed in Italy. She was told that this was a cyst, but no report of the histology was available. He did not have any small cysts on his chest, abdomen, back, axillae, or groins.

II.7 D.C. Female aged 62 years. She has never had any bowel symptoms and had not noticed any cysts on her skin, but clinical examination showed two small cysts on the front of her chest, identical to those on her son's chest. When these were pointed out to her, she said that they had been present since the age of 17 years, but had not bothered her in any way.

IV.1 M.C. Female aged 12 years. IV.2 M.C. Male aged 8 years. Neither of these children had bowel symptoms or cysts on the skin. Sigmoidoscopy has not been done.

**Histological findings**

**Bowel.** All three patients who had a colectomy showed large numbers of adenomatous polyps throughout the colon.

The colon from the proband, III.1, was studded with numerous small sessile tumours, but in the sigmoid colon there were about 20 larger pedunculated lesions (Fig. 5). Histological examination of all the lesions showed benign adenomatous polyps. Several of the polyps in his sister, III.2, showed local malignant change. At his first operation his father, II.1, had an adenocarcinoma of the rectum with colloid degeneration; at the second operation only benign adenomatous polyps were found.

**Cysts.** Histological specimens were available on both types of cyst in the proband. Of the six cysts on his...
head and limbs, only the one on his left thigh was sent for histological examination. After being stained with haematoxylin and eosin this showed a typical epidermoid cyst (Fig. 6). The lining of the cyst was composed of normal epidermis with a basal layer, three to four prickle cell layers and a pronounced granular layer. The content of the cyst was flaky, basophilic staining keratin.

Two cysts were removed from his chest. Each showed a thin-walled cyst with a wavy lining and sebaceous glands opening into the wall (Fig. 7). The changes were those of steatocystoma multiplex.

**Figure 5.** Proband (III.1). The sigmoid colon showed many small sessile polyps and several larger pedunculated tumours.

**Figure 6.** Histological examination. Wall of cyst removed from left thigh of proband showing typical epidermoid cyst. (H+E × 250.)

**Genetic findings**

II.1 seems to be the first recipient of a new mutation for Gardner's syndrome. His parents (I.1 and I.7) were both Italian, but unrelated. They had no bowel symptoms or skin cysts and died at the ages of 65 and 78 years. He had five sibs; two of these died in childhood of influenza and tuberculosis. The other three are alive and well but neither they nor their children have any bowel symptoms or skin cysts.

II.7 may be the recipient of a new mutation for steatocystoma multiplex. Her father, I.3, who was Italian, was killed in the first world war at the age of 32 years. Her mother, I.4, who was of French extraction, died at the age of 81 years, and it is almost certain that she was uninvolved because II.7 looked after her for many years before her death. There was no consanguinity in this family.

The children of the proband as yet show no stigmata of either disease, but they will have clinical and sigmoidoscopic examination at regular intervals from the age of 13 years. No medical information is available on the four children of III.2.

The proband and his sister have inherited Gardner's syndrome from their father, i.e. in an autosomal dominant manner. The steatocystoma multiplex is probably also inherited in the same way, because both mother and son were affected.

**Discussion**

Gardner's syndrome (Gardner, 1951) is usually described as a triad of soft tissue tumours, hard tissue tumours, and polyposis coli. The syndrome was differentiated from familial polyposis in a series of publications about a single family by Gardner and his co-workers from 1950 to 1969. The family was first described (Kindred 109; Gardner and Stephens, 1950) as polyposis coli, when a medical student noticed the high incidence of carcinoma of the bowel in the family. Several affected members were found to have skin tumours, but at this stage they were thought to be unrelated. A special clinic was set up to investigate this family and subsequent publications reported the results of these findings. It became apparent that the soft tissue and hard tissue tumours that occurred were only present in patients who also had the polyposis (Gardner and Richards, 1953) and this triad then became known as Gardner's syndrome. Later, desmoid tumours and abnormalities of the teeth were added as associated findings (Gardner, 1962). From the pedigree shown it was seen to be inherited as an autosomal dominant trait (Gardner, 1951).

The skin cysts which occur in Gardner's syndrome are epidermoid cysts (Leppard, 1974). These may be solitary or multiple, can occur anywhere on the body surface, but most commonly on the legs and face. They often appear first in childhood. They are usually described as being numerous (Cramer, 1962; Fader et al, 1962) and large and disfiguring (Weary et al, 1964; Thomas et al, 1968; Alm et al, 1973). This usually means that they are situated on the head and neck and are, therefore, obvious on clinical examination, but this is not always the case. Thirty per cent of Leppard
and Bussey’s patients (1975) had solitary cysts and the average number per patient was only four. They were only large and disfiguring in one of their 39 patients.

There has always been some confusion about the cysts in Gardner’s syndrome. They have been called a variety of names such as epidermoid inclusion cysts (Coli et al, 1970), epithelial cysts (Stritzler, 1963), sebaceous cysts (Marshall, Martin, and Mackay, 1967), and sebocystomatosis or steatocystoma multiplex (Oldfield, 1954; Gorlin and Chaudhry, 1960). In some instances the terms sebaceous cysts and epithelial inclusion cysts are used alternately as if they were the same condition (Kenny and O’Neill, 1958; Fader et al, 1962; Thomas et al, 1968) and in others, patients were said to have both types of cyst (Gardner, 1956; Pastinszky, Vér, and Bernát, 1969). Where the histological features of the lesion were described it was almost always that of an epidermoid cyst (Gardner and Richards, 1953; Kenny and O’Neill, 1958; Staley, 1961; Gardner, 1962; Weary et al, 1964; Dunning and Ibrahim, 1965), and the report by Leppard and Bussey (1975) confirmed that this was the only type of cyst that occurred in Gardner’s syndrome.

Steatocystoma multiplex is often used to describe any patient with multiple ‘sebaceous cysts’. Individual patients and families with this condition are described in the published reports as having large numbers of disfiguring cysts (Oldfield, 1954) and as having cysts widely scattered on the trunk, scrotum, and extremities (Weary et al, 1964; Rosten, 1972).

Steatocystoma multiplex produces a distinct clinical and histological picture. The lesions are small, skin coloured, or yellow or bluish nodules which are situated symmetrically on the chest, back, axillae, and groins (Pringle, 1899; Anderson, 1950). They are usually described as being smaller than a large pea. They appear in late adolescence or in early adult life and then remain static for the rest of the individual’s life. They are thought to be a form of hamartoma (Kligman and Kirschbaum, 1964). The cysts are lined by wavy, crenulated epithelium with an ultra-thin horny layer, and various adnexal structures can be found within the wall of the cyst or discharging their secretions into it. The most frequently occurring appendage is the sebaceous gland, but hair follicles and eccrine sweat ducts may also be seen.

This condition is also said to be inherited as an autosomal dominant trait and can thus be expected to affect approximately half of the children of an affected person in each generation.

The patient described here (III.1) showed both of these rare conditions. Gardner’s syndrome, evidenced by adenomatous polyps of the colon, multiple epidermoid cysts on the head and limbs, and desmoid tumours of the anterior abdominal wall, was inherited from his father. The steatocystoma multiplex, with very few though typical cysts on his chest and abdomen, was inherited from

Fig. 7. Histological features. Wall of cyst removed from chest of proband showing typical wavy lining of steatocystoma multiplex and a sebaceous gland within the wall of the cyst. (H & E × 90.)
his mother. Though his children show neither of these conditions at present, they will be kept under observation to see if either or both disorders develop in the future.

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REFERENCES


A giant short arm of no. 21 chromosome in mother of 21/21 translocation mongol

Summary. An extreme variation of the short arm of no. 21 chromosome in the mother of a 21/21 translocation mongol is described. The possible relation between the very long short arm of chromosome 21 in the mother and a centric fusion type of translocation mongolism in the offspring is discussed.

We recently encountered an interesting family where the mother had an abnormal no. 21 chromosome and the child had a centric fusion type of translocation Down's syndrome.

Case report

The patient was a 10-year-old boy with the typical features of Down's syndrome. The mother was 20 and the father was 25 years old at the time of his birth. The mother's phenotype was entirely normal.

Chromosome studies

The patient's chromosome analysis showed that he had 46 chromosomes with 21/21 centric fusion type translocation (or isochromosome of no. 21 chromosome). This cytogenetic finding was confirmed by trypsin banding technique (Fig. 1).

The father's karyotype was normal. The pheno-
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doi: 10.1136/jmg.13.5.407

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