GARDNER'S SYNDROME

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Gardner's syndrome consists of 3 different tumours in one patient: colonic polypi, osteomata of the skull and mandible, and benign subcutaneous tumours. It is a hereditary condition caused by a dominant defective gene.

In 1951 Gardner and Richards[1] reported 6 cases in 51 living relatives; 8 deceased members of this family had died from colonic carcinoma. Weiner,[2] Smith,[3] and Staley[4] added to the literature on this subject and stressed the danger of carcinoma developing in the polypi of the colon in contrast to Peutz-Jeghers syndrome (small bowel polypi with pigmented oral mucosa).

The soft-tissue tumours were subcutaneous cysts, lipomata and fibromata, with proliferation of fibrous tissue in scars—as was clearly shown in the case described below.

CASE REPORT

A White male, aged 29, developed lumps on his scalp and mandible and subcutaneous tumours on his limbs and trunk at an early age, but was otherwise fit and well in all respects. He presented himself for examination because, for a year, he noticed rectal bleeding on defaecation. He had no other symptoms.

On examination he was a very fit man generally. Rectally polypi were easily felt, and at sigmoidoscopy multiple polypi were seen, of which 2 were removed for histological examination.

The histological report (Dr. W. G. Davis) read as follows:

> 'The lesions have a papillary adenomatous structure. Almost all the epithelial cells are goblet cells, some of the glands are atypical, being abnormally large and irregular in shape. These glands are lined by several layers of cells. The epithelial cell nuclei show little pleomorphism, and mitoses are scanty. Between the glands is a loose connective-tissue stroma containing small round cells. Muscularis mucosae is present and this does not show invasion. The lesions are rectal polypi, and no clear-cut evidence of malignancy has been demonstrated.'

A barium enema (Dr. J. A. Beyers) showed polyposis of the transverse, descending, and especially the sigmoid colon.

Skull radiographs showed numerous osteomata of the skull and mandible.

The limbs and trunk showed multiple subcutaneous tumours of which several were excised; these were sebaceous cysts. The scars developed hypertrophic fibrous tissue.

At abdominal operation (Prof. F. du T. v. Zijl) a resection of the sigmoid colon and of the rectum was done. Enlarged mesenteric glands were found. The condition appeared to be malignant.

The pathologist at the University of Stellenbosch (Prof. H. W. Weber) reported on the resected specimen:

> 'The colon shows multiple polypi with loss of epithelial differentiation. No infiltration of the colon is present, but the changes in the epithelium should be regarded as a transitional phase between a benign and a malignant state. The lymph glands show no sign of malignancy.' (Translated from the Afrikaans.)

The patient has subsequently (2 years now) not reported any trouble.

On inquiry no definite family history of this condition could be elicited, but the patient's knowledge of his family was superficial.

He has two children aged 4 and 2, and the elder boy shows a number of osteomata developing on the skull. He will be watched.

Of additional interest was a family history of porphyria, but the patient's stool investigation was negative for porphyrins.

This is the first case of Gardner's syndrome reported in the South African literature.

The medical team responsible for the full investigation of this case to whom thanks are due are: Dr. Ben Swart, Oudtshoorn; Prof. F. du T. v. Zijl, surgeon, Cape Town; Dr. W. G. Davis, pathologist, Cape Town; Dr. André Beyers, radiologist, Cape Town; and Prof. H. Weber, Dept. of Pathology, University of Stellenbosch.

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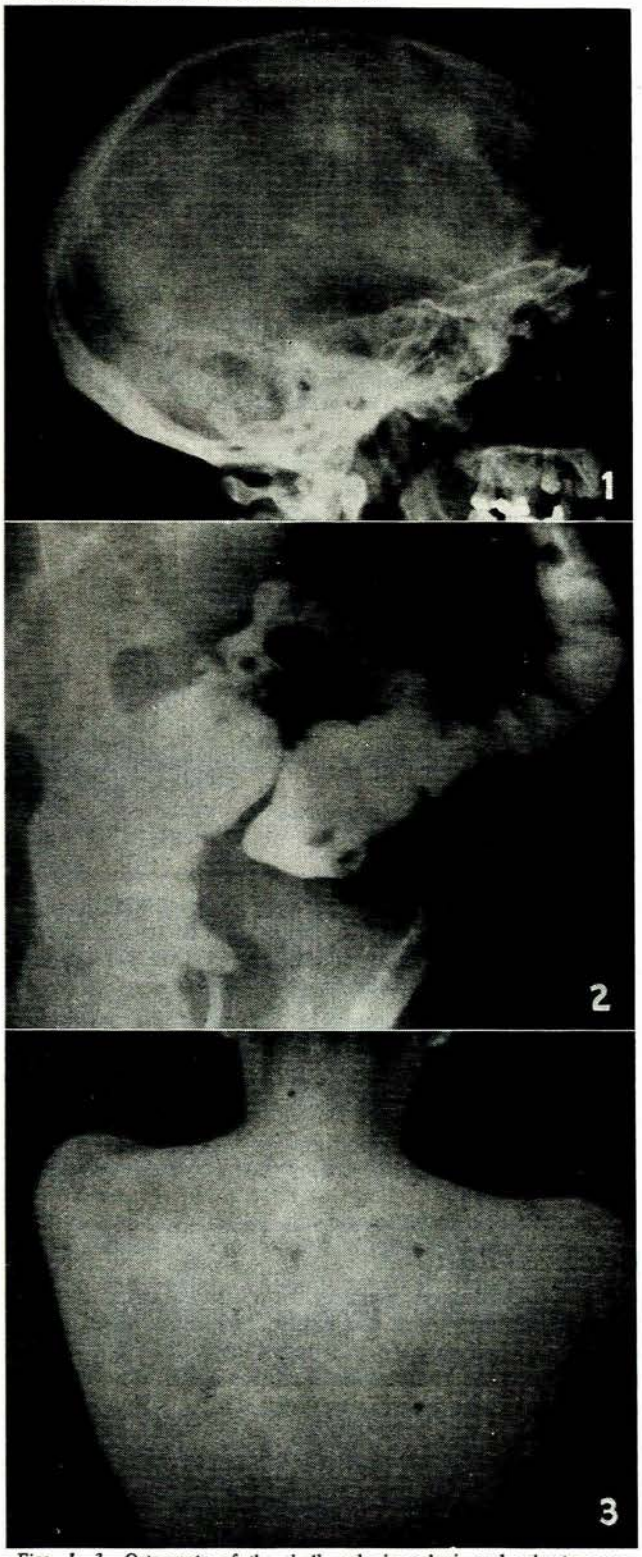

Figs. 1 - 3. Osteomata of the skull, colonic polypi, and subcutaneous tumours, respectively.