

THE GORLIN'S SYNDROME: A CASE REPORT

K. C. LING
N. T. A. COUPER
W. S. HU

SUMMARY

A relatively uncommon case of Gorlin's syndrome is reported. The jaw cysts led to the diagnosis of Gorlin's syndrome in the patient.

INTRODUCTION

Gorlin's syndrome, also known as multiple basal cell naevi syndrome, is a well documented disorder of unknown etiology. Very often it is familial of autosomal dominant mode of transmission. It can affect any age group of both sexes. It presents a wide spectrum of signs and symptoms involving nearly every system of the body. Classically it consists of triad features of multiple basal cell naevi, jaw cysts and rib anomalies. However, many other associated defects had been reported, including calcification of falx cerebri, bridging of sella turcica,

orofacial defects, fusion of cervical and thoracic vertebrae, shortened metacarpals, palmar and plantar pits, epidermal cysts and ocular anomalies.

The purpose of this paper is to highlight the significance and management of orofacial manifestations of this syndrome.

CASE REPORT

A 16-year-old Indian boy was referred to the senior author on 13 March 1981 by a general dental practitioner for management of a right mandibular swelling. The swelling was of three months duration. There was associated occasional discharge intraorally but no systemic upset.

He also gave a history of multiple nodules found on his body since the age of 11. They were painless but gradually increased in size and number. There were no other relevant medical history. The parents and other six siblings were not similarly affected.

The patient was treated for a painful right nasolabial swelling when he was 12-years-old. It was suspected to be an infected cyst and subsided after a course of antibiotics. Patient defaulted subsequent follow-up.

Physical examination found him in good health. He was rather tall — 175 cm in height — for his age. 24 painless soft tissue swellings were found on different parts of the body like the neck, upper

K. C. Ling, BDS, MDS, FDSRCS
Lecturer, Department of Oral Surgery
Faculty of Dentistry
University of Malaya
Kuala Lumpur

N. T. A. Couper, BA, MBBChir, DCH, FRCR
Lecturer, Department of Radiology
Faculty of Medicine
University of Malaya
Kuala Lumpur

W. S. Hu, MBBS
Medical Officer, Department of Radiology
University Hospital
Kuala Lumpur

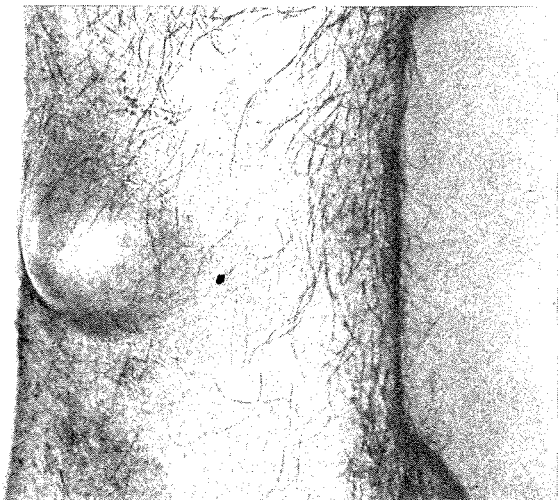


Fig. 1 A large epidermal cyst on the lateral aspect above the ankle of the right leg.

limbs, abdomen, back of trunk and legs (Fig. 1). They varied from 0.8 cm to 2.5 cm in diameter. They were confirmed by the general surgeon as epidermal cysts. There were no basal cell naevi on his body.

Extraoral examination showed an obvious non-tender bony-hard swelling at the right angle of the mandible. It extended up the lateral surface of the ramus. The lower border of the mandible below the right first and second molars was also expanded. There was no right labial anaesthesia indicating that the right inferior dental nerve had not been affected by the lesion. Right upper cervical lymph nodes were palpable. He had a rather broad nasal root.

On intraoral examination, a bony mandibular swelling extending from the right second premolar backward and upward to the coronoid process of the same side was observed. It was non-tender. No sinus was seen. The adjacent teeth were sound and firm. Only the right lower third molar was missing. The palatal vault was rather high.

Radiological examination showed a lytic lesion occupying the right angle and almost the whole right ramus of the mandible (Fig. 2). Only the condyle was not affected. Within the lesion there



Fig. 2 Orthopantomogram showing a lytic lesion occupying the angle and much of the right ramus of the mandible. Note the grossly displaced third molar.



Fig. 3 Pan-oral radiograph showing the three maxillary lytic lesions. Note also the abnormally constricted neck and curved roots of the left second molar.

was the clinically missing third molar which had been grossly displaced. A smaller lytic lesion was also seen at each of the following three sites in the maxillae (Fig. 3): between the lateral incisor and the canine of the right side; around the root apex of the lateral incisor of the left side; around the root apices of the first molar of the left side. The left upper second molar also had an abnormal tooth morphology in that the neck was much constricted and the roots curved (Fig. 3). A provisional diagnosis of multiple jaw cysts in a Gorlin's syndrome was made.

Under general anaesthetic the three maxillary lesions were enucleated. The mandibular lesion was marsupialised in view of its large size and the cavity packed with medicated gauze. The pack was removed six days later and the opening kept patent by an obturator. About seven months after the marsupialization, the mandibular lesion, which was much smaller in size, was enucleated. All the removed

maxillary and mandibular lesions were sent for histological examination.

The histological examination of all the four lesions showed them to be odontogenic keratocysts. The haematological and bio-chemical investigations gave results within normal limits. A skeletal survey done showed the following defects: bridging of the sella turcica; calcification of the falx cerebri (Fig. 4); rib anomalies which include bifid third ribs of both sides and posterior splitting of second and fourth ribs of the left side (Fig. 5); shortened fourth and fifth metacarpals of both hands (Fig. 6). A final diagnosis of Gorlin's syndrome was then made.

On follow-up, a recurrent cyst was found between the lateral incisor and canine of the right maxilla about two years after the surgical treatment. A new cyst was discovered around the left lower impacted third molar at about the same time. Both cysts were enucleated under local anaesthetic.

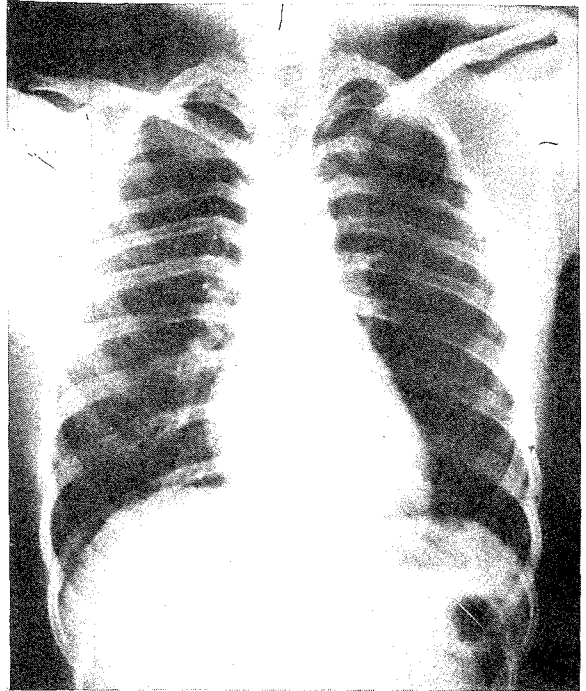


Fig 5 Postero-anterior chest x-ray showing the bifid third ribs of both sides and posterior splitting of the second and fourth ribs of the left side.

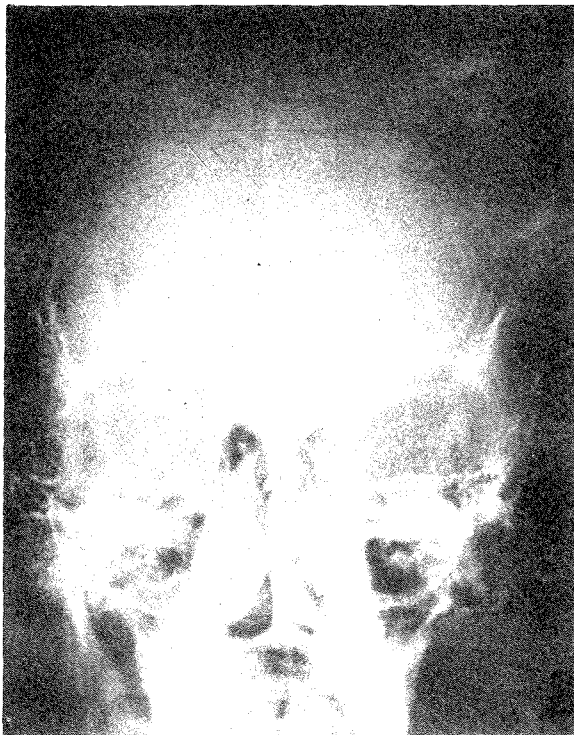


Fig. 4 Postero-anterior skull radiograph. The lamellar calcification of the falx cerebri is an abnormal feature in a 16-year-old.

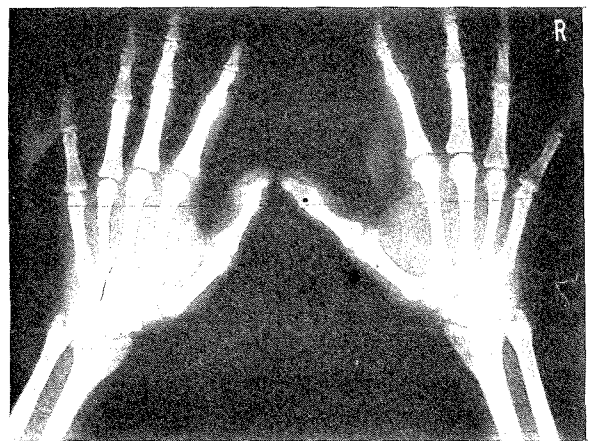


Fig 6 Antero-posterior x-rays of both hands showing the shortened fourth and fifth metacarpals.

DISCUSSION

The diagnosis of Gorlin's syndrome can be firmly established if the patient has at least naevi or jaw cysts combined with either a calcified falx cerebri

or palmar or plantar pits.¹ This patient had multiple jaw cysts, calcified falx cerebri and other associated defects.

The reported orofacial defects of Gorlin's syndrome were frontal bossing, ocular hypertelorism, broad nasal root, mild mandibular prognathism, multiple fibroepithelial polyps of the tongue, high palatal vault, multiple jaw cysts and dental defects like geminated and missing teeth. This patient has broad nasal root, high palatal vault, multiple jaw cysts and dental defects.

The reported jaw cysts, which were almost always odontogenic keratocysts, developed early in life. They were the usual presenting symptoms of the syndrome. This is significant as they could lead to a diagnosis of Gorlin's syndrome. This in turn helps in the early detection of related patients who may be similarly affected.

There are three generally accepted lines of treatment for the jaw cysts, enucleation, marsupialization and a two-stage plan where marsupialization was followed by enucleation after a delay period of several months. In this patient both enucleation and two-stage plan were carried out.

The surgically treated jaw cysts had great propensity to recur. The incidence of recurrence by a single cyst was about 25%.^{2,3} When the recurrence is related to the patient the rate was 85%.³

In a reported series of six patients the average time elapsing before recurrence varied from two to nine years.² In this patient the time elapsing before recurrence was two years. It is believed that epithelial islands and microcysts located in the connective tissue between the oral epithelium and the cyst wall may be one explanation of the tendency of these cysts to recur.⁴

New jaw cyst can continue to develop through life, although with a much reduced frequency. In this patient a new cyst developed during follow-up.

Ameloblastic and sarcomatous changes had been reported in the jaw cysts.

The reported dental defects were few. In this patient the defect was the abnormally constricted neck and curved roots of the left upper second molar. The significance of this defect is that ordinary forceps extraction of this tooth would probably result in a fracture of either the crown or roots. This tooth, if it requires removal, should be removed by a surgical procedure. To avoid possible complications from abnormal tooth morphology during extraction, patients with Gorlin's syndrome should have all the teeth x-rayed.

The basal cell naevi in Gorlin's syndrome usually appear when the patients are in their 20s' or 30s'. Thus it is not surprising that this patient had no such naevi. The basal cell naevi, when present, tend to become basal cell carcinomas.

In view of the clinical behaviour of the jaw cysts and the high risk of developing basal cell carcinomas later in life, it is essential to closely follow-up patients with Gorlin's syndrome for as long as possible, if not life. This would ensure early detection of basal cell carcinomas. The jaws should be x-rayed yearly to detect any recurrence or new cyst and if detected could be adequately removed because they are small.

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