Medical Memoranda

Hereditary Gingival Fibromatosis

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Hereditary gingival fibromatosis is an uncommon disorder. Its clinical features are variable, and in the following case the patient showed some hitherto unreported abnormalities.

Case Report

The patient, an 18-year-old girl, was born at full term after an uneventful pregnancy, her birth weight being 2 kg. She was slow in reaching her milestones in childhood and has always been mentally retarded. At the age of 13 she developed hirsuties of the face, limbs, and back, and this became progressively more severe. Her menarche was at 13, since when her periods have been irregular, occurring every two to three months, and lasting up to six or seven days.

When first seen at the Royal Victoria Infirmary by Professor G. A. Smart her height was 171 cm., span 142 cm., and crown-to-pubis measurement 71 cm. Her weight was 40·6 kg. There were coarse dark terminal hairs on the cheeks, chin, forehead, arms, legs, lower back, and areolae. The eyebrows were bushy, but the scalp hair was normal and the pubic hair was of the female distribution. There was a little acne of the face with a disproportionate seborrhoea. The skin felt doughy and was hyperelastic and she had more than 50 benign pigmented cellular naevi widespread over the body. The breasts were poorly developed and there was no clitoral hypertrophy. The internal genitalia were also normal.

The gingival mucous membrane, particularly of the upper jaw, was excessive, giving the palate a deep and narrow appearance (Fig. 1); many teeth were partially buried by this hypertrophic tissue. There was also an increase in the size of the teeth-bearing part of the bony maxilla. Apart from obvious mental deficiency other systems were normal.

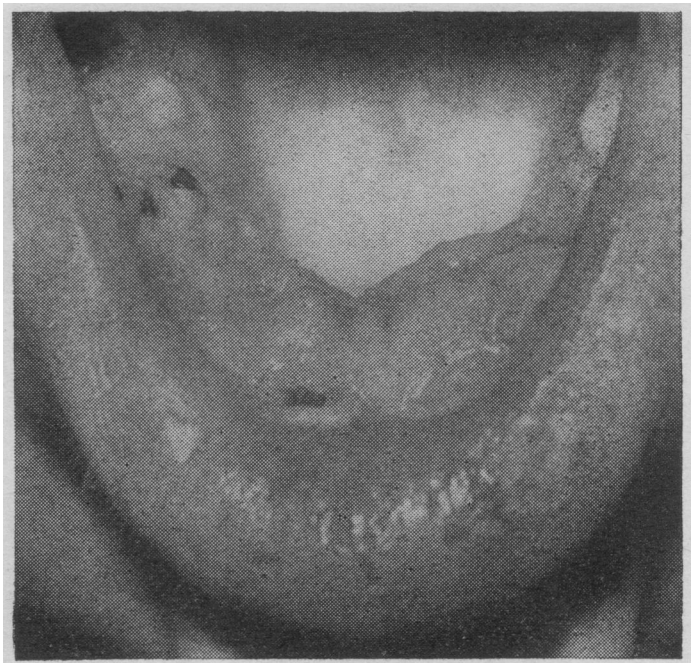

FIG. 1.—Hypertrophied gingival mucous membrane.

Investigations

Haematological, biochemical, and radiological investigations are shown in the Table. An unexpected abnormality was found on x-ray examination of the cervical spine. Dr. A. Appleby reported: "there is a curious anomaly of the second cervical spinous process, which is very narrow. In addition the odontoid is unusually long and reaches almost to the base of the clivus. The first ribs arise from the eighth vertebra, which, however, has the shape of a cervical vertebra. The first ribs themselves are abnormal and articulate with the second rib on each side" (Fig. 2).

A gingival biopsy showed an increase in the submucosal fibrous tissue compatible with the diagnosis of hereditary gingival fibromatosis. A biopsy of the skin, including "elastic" stains and stains for mucopolysaccharides, showed no abnormality. Total skin collagen was normal (232 μg./sq. mm. of skin surface).

Dermatoglyphic studies showed that the overall digital ridge counts were in no way aberrant from those of her first-degree

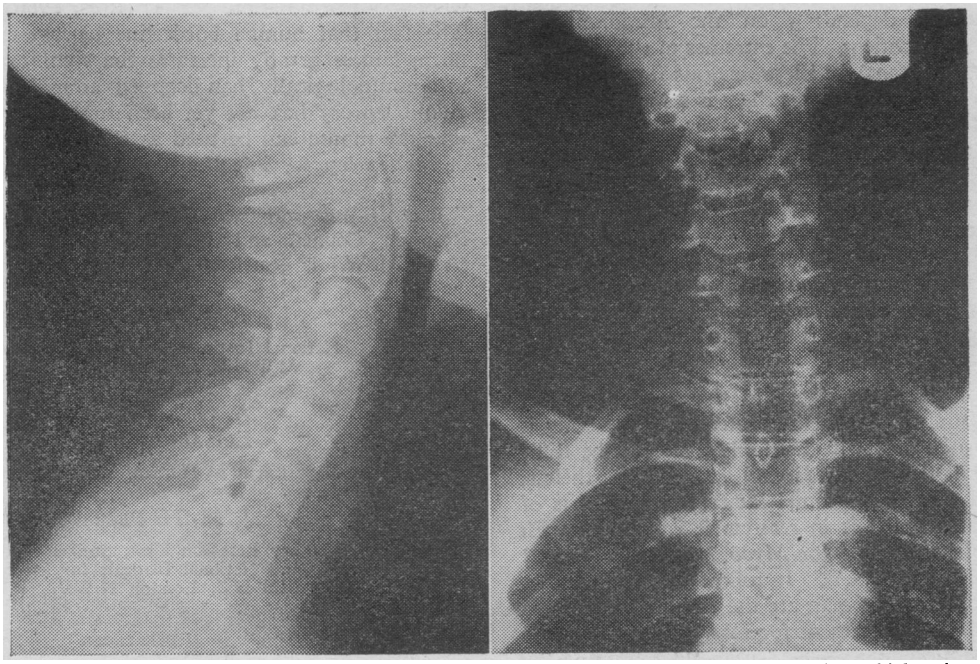

FIG. 2.—Unusually long odontoid process, narrow second cervical process, and first ribs which arise from the eighth vertebra, which has the characteristics of a cervical vertebra. The first ribs also articulate with the second ribs.

relatives, the only unusual feature being the three triradii associated with a twin loop on the left thumb. On the palms the b-c ridge count was considerably raised on both hands as compared with her first-degree relatives and beyond the range seen in 3,000 normal schoolchildren (Roberts and Coope, 1969). A full-scale I.Q. was 50 and an E.E.G. showed a moderately non-specific abnormality with a generalized excess of slow activity.

Summary of Investigations

Investigations	Results
Haematological	Haemoglobin 13·2 g./100 ml., W.C.C. 6,700/cu. mm., blood group A positive
Biochemistry:	
Blood urea and electrolytes	19 mg./100 ml., electrolytes normal
Serum proteins	7·1 g./100 ml., electrophoresis normal
Mucopolysaccharides (Mansley–Hawksworth test)	Not detected
Urinary amino-acid chromatogram; urinary hydroxyproline	Normal
Routine urine	No albumin or reducing substances
Endocrine studies:	
Urinary 17-ketosteroids	11·7 mg./24 hours
Urinary 17-hydroxycorticosteroids	4·8 mg/24 hours
Urinary 11-hydroxycorticosteroids	141 μg./24 hours
Plasma 11-hydroxycorticosteroids	Normal diurnal variation from 19·5 μg./100 ml. at 9 a.m. to 7·4 μg./100 ml. at midnight
Protein-bound iodine	7·1 μg./100 ml.
Radiological:	
Chest	No abnormality in lung fields or heart
Abdomen, skull (including maxillae), and skeletal survey	Normal, bone age 17
Cervical spine	See text and Fig. 2

Genetic Study

Fig. 3 shows the pedigree of the proposita, the eldest of four surviving children. The two abortions were not related to the condition. There is no consanguinity in the family. Physical examination of both parents showed them to be unaffected, and there was no report of any other case in the family on either side.

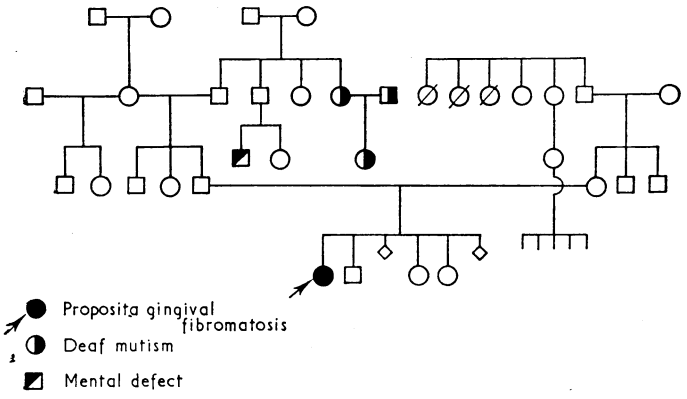

FIG. 3.—Pedigree of proposita.

The patient's father's father's sister is a deaf-mute who married a deaf-mute and has a deaf-mute daughter. It seems that this is a recessive form of deaf-mutism, and there is no evidence that this is in any way associated with the condition in the proposita. The proposita's father's father's brother's son is a mental defective, but nothing further is known of him. It was impossible to investigate members of this side of the family since they live overseas.

In view of the fact that hereditary gingival fibromatosis is a well-established dominant, and is not of low penetrance, the pedigree was interpreted as indicating that the proposita's condition was the result of a new mutation.

Comment

Hereditary gingival fibromatosis is a rare condition. Its mode of transmission is usually a dominant trait, and occasionally, as in the present case, it is the result of a new mutation. Clinically it is characterized by a firm, painless enlargement of the gums. The hyperplastic tissue is of normal colour and not especially liable to trauma and the degree of swelling varies from the mild to the grotesque. The enlargement usually begins with the eruption of the permanent dentition, occasionally with the eruption of the deciduous dentition, and rarely is present at birth. A few cases have arisen in adult life (Rushton, 1957). Frequently the hyperplasia is the only abnormal finding, but other defects may be associated. Hypertrichosis is the most commonly associated feature ; this may be present at birth (Byars and Sarnat, 1944), but, as in our patient, it often appears at puberty. The age of onset of hypertrichosis is unrelated to the age of onset of the gingival fibromatosis. Mental retardation is the second most frequent association.

Epilepsy (Weski, 1920 ; Ramon *et al.*, 1967), defective limb appendages, large ears, and nasal abnormalities are infrequent features of this syndrome and were not present in this patient. She, however, showed other hitherto unrecorded features. She had certain cutaneous features which should be looked for in other patients, the skin felt doughy, was hyperelastic, there was gross seborrhoea, and she had many benign pigmented cellular naevi. *X*-ray examination of the cervical spine also showed an unusual second cervical vertebra, an abnormality which has not previously been reported in this syndrome.

This syndrome should therefore always be considered as a diagnostic possibility when patients present with "idiopathic hirsuties," epilepsy, or mental retardation, and a search should be made for gingival hyperplasia and for other associated defects in such patients.

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