Clinical records

Focal dermal hypoplasia

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Focal Dermal Hypoplasia is a rare hereditary syndrome which has never been reported in the otolaryngological literature. Gorlin *et al.* (1963) described two cases and reviewed eleven other reports of the same condition. An additional case has since been reported by Dugois *et al.* (1965). As many of the characteristic lesions of this syndrome are found in the head and neck region it was considered that the following case report would be of interest for otolaryngologists.

Case report

A 34-year-old white female was referred to the Department of Otolaryngology in November 1969 for removal of a wart-like tumour from the right nasal vestibule. She was one of three living children, a fourth pregnancy having resulted in a male stillbirth. There was no history of parental consanguinity. Family history was negative apart from a cousin of the patient who had syndactyly. The patients' pregnancy and delivery were uncomplicated but syndactyly of the second and third digits of the right hand and left foot, and red, papular lesions of the skin of limbs and trunk were found at birth. Lesions around the vulva and anus appeared at age seven and were diagnosed as condylomata acuminata. An adenotonsillectomy and right radical mastoidectomy were performed at eighteen years of age for a chronic suppurative otitis media.

The patient was of normal intelligence. There was a slight prognathism and the vermilion borders of the lips showed several small verrucous growths and numerous pinpoint telangiectases. The upper jaw was edentulous but the lower incisors were present, were irregular in size and form and showed enamel hypoplasia. A 4 mm. cleft of the tip of the tongue was present and there was a solitary papilloma on the dorsum just anterior to the foramen caecum. The tonsil beds were well healed but the operation report from 1954 describes the tonsils as having an 'unusual papillomatous appearance'. Unfortunately histological examination was not performed.

Nasal examination revealed notching of the left ala nasi and the papilloma of the right nasal vestibule for which she had originally been referred.

The external ears were large, closely applied to the head and lacking the normal degree of rigidity, the cartilage of the pinna being uniformly thin bilaterally. A papilloma was present in the floor of the right cartilagenous meatus. The left drumhead was of normal appearance and on the right side there was a radical mastoid cavity. A pure-tone audiogram showed normal hearing in the left ear and a 70 dB combined loss in the right.

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Ophthalmic examination revealed a convergent strabismus of the right eye and bilateral colobomas of iris and choroid. The nails of all digits were thin and brittle. On the trunk and extensor surfaces of the limbs there were numerous areas of thin, atrophic skin, yellow-red in colour, which ballooned irregularly above the level of the epidermis. These areas could be easily reduced into the deeper tissues and gave the impression of herniations through the dermis.

Routine haematological, serological and urine investigations were non-contributory. Chromosome analysis was normal. A skeletal survey was unremarkable apart from a shortening of the right fourth metacarpal. An intravenous pyelogram and double-contrast barium enema did not reveal any abnormalities and oesophagoscopy and laryngoscopy were negative.

Excision biopsies of the lip, oral, nasal and aural tumours showed the typical appearance of papillomata. Histology of a representative skin area (Fig. 1) showed an epidermis of almost normal thickness with moderate acanthosis, small rete pegs and moderate cornification. Deep to the epithelium there was a very thin layer of dermis interposed between the epidermis and a broad layer of mature adipose tissue. In several areas the subcutaneous fat extended almost to the undersurface of the epidermis.

The patient was seen again in January 1970 when she presented with a large papilloma of the lower lip (Fig. 2) which had attained a diameter of 1 cm. in the course of two months. There was no recurrence in the ear canal or nasal vestibule.

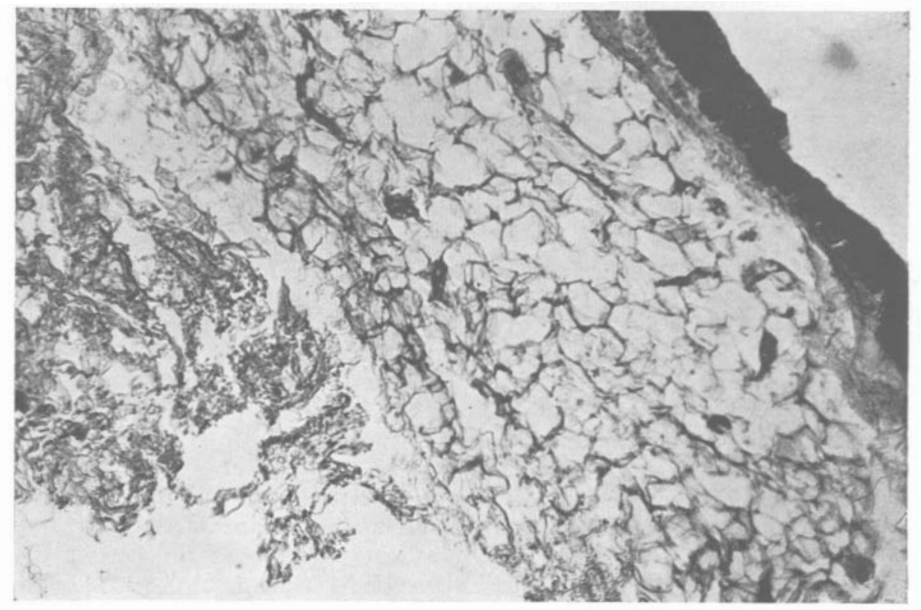


FIG. 1.

Representative skin lesion showing a somewhat darkly-staining epidermis, a very thin dermis and a broad layer of mature adipose tissue which at one point almost touches the undersurface of the epidermis. Haematoxylin and eosin, × 72.

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FIG. 2.
Papilloma of lower lip.

Discussion

Focal Dermal Hypoplasia appears to be transmitted as an autosomal dominant with incomplete penetrance. Most of the reported cases have been female and similar defects have been found only in female relatives. There is a high incidence of miscarriages in these families and it is postulated that the condition is incompatible with survival in the male foetus (Goltz *et al.*, 1962). Chromosome abnormalities have not been demonstrated (Gorlin *et al.*, 1963).

Papillomas of the lips have been a striking feature of several reported cases (Naegeli, 1927; Dugois *et al.*, 1965) and one of the three cases reported by Goltz *et al.* (1962) had three recurrences subsequent to attempts at surgical removal. The growth potential of these papillomata is well demonstrated by the case reported above. Papillomata of the oral mucous membrane have been described only twice in the literature (Naegeli, 1927; Liebermann, 1935). Gorlin *et al.* (1963) have also reported a cleft of the tongue and the same authors described a case with double frenulum of the upper lip. Dental abnormalities have been more frequently reported with microdontia (Cole *et al.*, 1941) and enamel hypoplasia (Wodniansky, 1957) the most frequent findings.

Notching of the ala nasi has been described by Gold (1958) and asymmetry of the nasal cartilages by Wodniansky (1957) but no other report of nasal papilloma could be found.

Abnormal positioning of the ears has been reported by Gorlin *et al.* (1963) and thinness of the pinna cartilage by Mannkopf and Hanney (1957). There is

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only one other report of papilloma of the external ear (Jessner, 1921). In view of the known association between deformities of the external ear and malformations of the genito-urinary tract (Hilson, 1957) and the resemblance between the large, flabby ears of our patient and those first described by Potter (1946), an intravenous pyelogram was performed but with negative findings. Middle and inner ear abnormalities have not been reported.

The most characteristic feature, however, of this syndrome is the skin abnormality and diagnosis can be established by the histological findings of a very thin dermis with focal defects and herniation of the subcutaneous fat through these defects.

Summary

Focal Dermal Hypoplasia is a rare hereditary syndrome of which there are only fifteen reports in the literature. A case is presented and the literature reviewed with special emphasis on the ear, nose and throat manifestations.

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