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Dyskeratosis congenita and nasopharyngeal atresia

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Abstract
Dyskeratosis congenita is a multisystem disorder with an increased incidence of neoplasia and opportunistic infections. A case is reported as a cause of complete nasopharyngeal atresia.

Case report

A 68-year-old retired coal worker presented in 1985 with pneumonia and progressive dyspnoea. He smoked twenty cigarettes daily and used a salbutamol inhaler and oxygen. For as long as he could remember he had been totally unable to breathe through his nose. He did not relate this to a previous nasal fracture or childhood tonsillectomy. There was no relevant family history.

Anterior rhinoscopy was unremarkable but on attempting posterior rhinoscopy a complete nasopharyngeal atresia was found with the soft palate in continuity with the posterior pharyngeal wall (Fig. 1). Rhinomanometry revealed no airflow or pressure change at all. The tongue and oral mucosa were red, smooth and atrophic and the tonsils were absent but without any visible abnormal scarring.

General physical examination revealed multiple grey pigmented reticular lesions over his back and abdomen. His finger nails and several toe nails were absent and had been since his teens (Fig. 2). Bilateral ectropion, periorbital oedema and chronic blepharitis were present (Fig. 3). His eye lashes were absent and bilateral cataracts noted. His chest was over inflated with quiet breath sounds, expiratory rhonchi and bilateral basal coarse crackles. Bronchial breathing was audible at the right apex. There was marked hepatomegaly.

His haemoglobin was 10 gms/dl with ferritin 11 µg/l. B12, folate and marrow cellularity were normal. Routine biochemistry, immunoglobulins, anti-nuclear factor, autoantibodies, angiotensin converting enzyme and aspergillus and avian precipitins were normal or negative. A chest radiograph showed an irregular, 2 cm right lower zone mass with faint generalized reticular shadowing and bilateral upper zone fibrosis.

Fibreoptic nasendoscopy confirmed complete atresia of the nasopharyngeal opening, the soft palate being totally fused with the posterior pharyngeal wall. There was however a post nasal space present which was of normal dimensions except for the pharyngeal opening which did not exist. At palatoplasty the area of fusion was found to be 1.2 cm in vertical length and 3 cm wide. It did not appear to be a concentric closing of the opening but an anterior to posterior fusion. The fusion was divided by sharp dissection and a silastic stent sutured to the soft palate. This was tolerated by the patient for four weeks then removed. Four months later the palate had re-stenosed.

He was followed up for two years thereafter until he died following the development of pneumonia. Post-mortem revealed a primary bronchial adenocarcinoma with severe emphysema and bilateral upper lobe fibrosis.

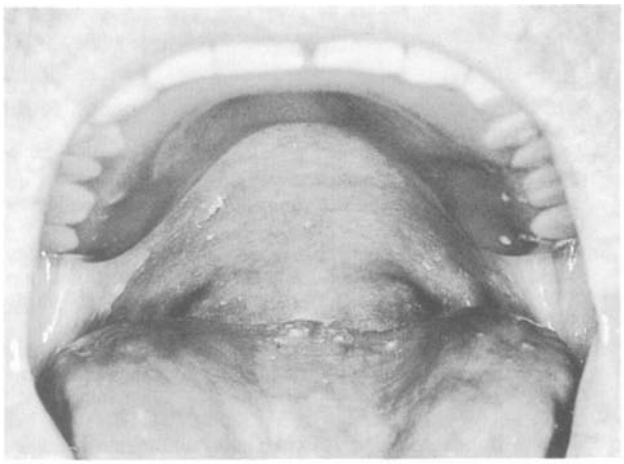

Fig. 1
Smooth and atrophic tongue and oral mucosa with complete atresia of the nasopharynx.

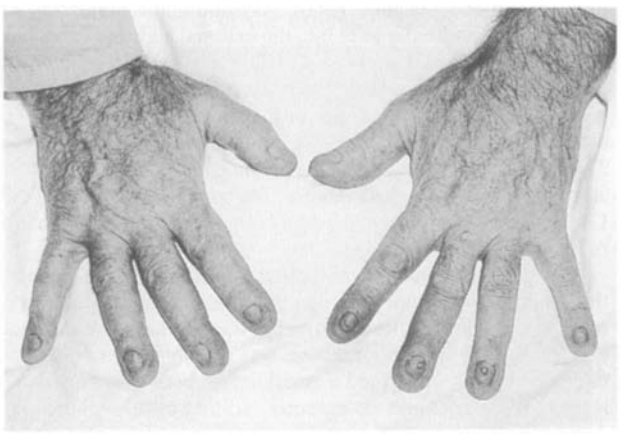

Fig. 2
Absent finger nails.

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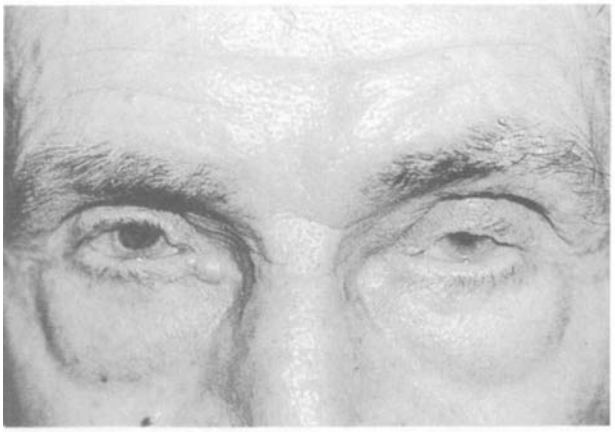

Fig. 3
Bilateral ectropion, periorbital oedema and chronic blepharitis.

Discussion

Dyskeratosis congenita is a rare, multisystem disorder, first described in 1910 by Zinsser. The classical triad is of reticulate skin hyperpigmentation, dystrophic nails and leukoplakia (Cole *et al.*, 1957). It is associated with increased malignancy (Sirinavin and Trowbridge, 1975). Approximately 80 cases have been described. The onset is generally before puberty (Trowbridge *et al.*, 1977). It is 10 to 13 times more common in males and affects mainly whites (Sirinavin and Trowbridge, 1975). Inheritance is usually X-linked but an autosomal dominant pattern also occurs. The gene has been localized to locus Xq28 (Connor *et al.*, 1986). Abnormal chromosomal breakage has been reported in one third (Connor and Teague, 1981). The underlying defect is uncertain.

Hyperpigmentation and nail dystrophy occur in virtually all cases and leukoplakia in 87 per cent (Sirinavin and Trowbridge, 1975). Hyperpigmentation usually affects the neck and upper chest (Cannell, 1971). Leukoplakia most frequently affects the oral mucosa but can occur in any mucous membrane. Dysphagia is present in 59 per cent and has been associated with subsequent malignancy. Oesophageal diverticulum, congenital oesophageal stenosis and post cricoid webbing have been described (Sirinavin and Trowbridge, 1975). Abnormalities of the external ear, transparent tympanic membrane, meatal atresia and congenital malformations of the middle ear have been reported (Ogden *et al.*, 1988). Eyes, teeth or hair are affected in most patients. In 48 per cent the build is hyposthenic and 42 per cent have subnormal intelligence. Endocrine, genital and skeletal abnormalities occur.

Haematological manifestations resembling Fanconi's anaemia occur in 58 per cent with anaemia and pancytopenia although the two conditions are distinct (Sirinavin and Trowbridge, 1975). The marrow is usually hypocellular. The commonest cause of death is infection, often opportunistic. Management is principally supportive although bone marrow transplantation may be considered.

There is an increased incidence of neoplasia with 16 per cent developing tumours usually squamous and mucosal principally in the mouth, tongue, nasopharynx and rectum (Sirinavin and Trowbridge, 1975). Squamous carcinoma of the bronchus has been described in one patient with associated rectal adenocarcinoma and distinct squamous carcinomas of the nasopharynx and tongue (Cole *et al.*, 1957). There are no previous reports of primary adenocarcinoma in the lung.

Nasopharyngeal atresia or stenosis has not been described in association with dyskeratosis congenita. Only 300 cases of nasopharyngeal stenosis have been reported worldwide. When acquired it is most often the result of adenotonsillectomy. In these cases there is still a lumen between the nasopharynx and oropharynx and complete atresia, as in our patient, has not been reported (Bennhoff, 1979). Complete nasopharyngeal atresia is reported as a further manifestation of this multisystem disorder.

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