Xeroderma Pigmentosum: Squamous Cell Carcinoma of the Tongue

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A case report is presented of a 9-year-old boy with xeroderma pigmentosum complementation group C, who died from a squamous cell carcinoma arising from the tip of the tongue. Treatment with etretinate and in a later phase with indomethacin and prednisolone was not effective, except for a limited initial response of the tumour to etretinate.

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Xeroderma pigmentosum (XP) is a rare autosomal recessive disease characterized clinically by cutaneous photosensitivity and pigmentary changes, photophobia and a propensity for the early development of malignancies in sun-exposed mucocutaneous and ocular structures (1). Cell biologic investigations have disclosed cellular hypersensitivity to UV radiation and certain chemicals in association with abnormal DNA repair. XP comprises at least nine excision defective complementation groups (A–I) and an excision proficient XP variant group.

In 13 XP patients, squamous cell carcinoma has been reported on the tip of the tongue (1–6). We describe an additional case: a 9-year-old boy with XP complementation group C. Treatment of the tumour with an oral retinoid (etretinate) gave only a limited initial response. No response was seen of treatment with indomethacin and prednisolone, as opposed to the experience of others (7).

CASE REPORT

The patient, a 9-year-old boy, was born in 1977 of non-consanguineous parents. His mother had two brothers (JW, DW) and one sister with XP complemantation group C (JW: XP 16 RO). He had photophobia and recurrent conjunctivitis and developed intensive freckling on sun-exposed sites. Cell biologic studies in 1980 according to methods previously described (8) showed abnormal unscheduled DNA synthesis after UV radiation (5–10% of the control level) of the patient's fibroblasts (XP 40 RO). From his fifth year the patient had several solar keratoses. In November 1985 a keratoacanthoma developed on the left temple, which disappeared with adjuvant treatment with etretinate. In July 1986 he presented with an erosive tumour at the tip of his tongue.

Dermatologic examination revealed hyperpigmented freckling diffusely spread over sun-exposed areas on the face, neck and the back of the hands. In addition, two solar keratoses were located on right cheek and nose. The dorsal surface of the tip of the tongue showed an indurated verrucous tumour (3.5×2.5×2 cm on palpation) with a small central ulceration. No regional lymphadenopathy was detectable.

Histologic examination (incision biopsies) of the tumour of the tongue showed an invasive well differentiated squamous cell carcinoma.

Aggressive surgical therapy aimed at total tumour excision (subtotal glossectomy), which would be very mutilating, was refused by the parents. Therefore tentative therapy with an oral retinoid (etretinate: 0.9 mg/kg) was started in August 1986. Initially the tumour showed some regression. Ten weeks after starting etretinate, however, the tumour had increased in size again. Etretinate therapy was discontinued. In October 1986 indomethacin (50 mg twice daily) and prednisolone (10 mg daily) were started and given for 4 months. No improvement whatsoever was seen. In the terminal stage, serious swallowing problems occurred and fever, weight loss and enlarged submandibular lymph nodes were observed. The boy died in February 1987.

DISCUSSION

Squamous cell carcinoma of the tongue is extremely uncommon in juvenile patients. The peak incidence of this neoplasm occurs between the sixth and eighth decade. The tumour is frequently associated with chronic alcohol and/or tobacco consumption. Approximately 20% of the squamous cell carcinomas of the tongue in non-XP patients are located on the anterior third, not on the tip but on the lateral edge of the tongue (10). Of 32 XP patients with lesions in the oral cavity, 16 had a squamous cell carcinoma, 13 of which were located on the tip of the tongue (1). Kraemer et al. determined the increase of the frequency of squamous cell carcinoma of the tip of the tongue of XP patients as more than 20,000-fold compared with a Caucasian control population (1). UV

radiation may play a role in the onset of this tumour. Our patient, who had never used tobacco or alcohol, presented with a tongue carcinoma even before cutaneous malignancies developed, as described also by others (6).

Malignant neoplasms on the anterior third of the tongue can be cured by prompt surgical excision in an early stage. Frazell & Lucas found a 5-year survival of 45.8% in patients with squamous cell carcinoma of the anterior third of the tongue (10). Because in our case radical excision of the tumour was refused, therapy with an oral retinoid (etretinate) was given tentatively. Treatment of advanced squamous cell carcinomas of the skin with isotretinoin has been described to be moderately effective (9). Initially the tongue tumour showed regression during therapy with etretinate. This regression might have been due to factors other than retinoid treatment, such as less inflammation, because of better hygienic care.

Therapy with indomethacin and prednisolone has been described to be effective for cutaneous squamous cell carcinomas in XP patients (7). In a group of 9 XP patients with squamous cell carcinoma of the skin, 3 patients showed complete regression of the tumour and 5 partial regression. One patient did not respond to treatment. In our patient too the squamous cell carcinoma of the tongue did not respond to this therapeutic approach.

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Familial Sarcoidosis: High Ethnic Prevalence

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We report three sisters of Irish extraction who all developed sarcoidosis. The cases emphasize both the familial and ethnic preponderance of sarcoidosis which has not been adequately emphasized in the dermatology literature.

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CASE REPORTS

Sister 1

Presented in 1961, with lethargy, enlarged axillary and cervical lymph nodes and chest radiograph showing bilateral hilar lymphadenopathy. She was diagnosed as having sarcoidosis and over the next few months the lymphadenopathy resolved spontaneously. Further details were not available as her hospital records had been destroyed.

Sister 2

Presented in 1980, aged 56, with a one-year history of an asymptomatic, 3 cm, purplish plaque on the left cheek. She