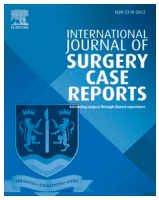
International Journal of Surgery Case Reports 88 (2021) 106513

Contents lists available at ScienceDirect

International Journal of Surgery Case Reports

journal homepage: www.elsevier.com/locate/ijscr



Case report

First report of oral angiokeratoma in a xeroderma pigmentosum

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ARTICLE INFO

Keywords:
Xeroderma pigmentosum
Angiokeratoma
Squamous cell carcinoma
Pyogenic granuloma
Basal cell carcinoma

ABSTRACT

Introduction and importance: Xeroderma pigmentosum is an autosomal recessive genetic disorder with impaired ability to repair DNA damage. Detection of the intraoral benign and malignant lesion in Xeroderma pigmentosum is uncommon, and the reported few cases were in the lip and tip of the tongue which are related to ultraviolet light exposure.

Case presentation: We present a case of 20 years old male with erythematous to violet, painless and shiny papule at the right side of the tongue. The lesion was surgically excised while the histopathological examination of the lesion showed focally eroded and hyperplastic stratified squamous epithelial covering with underlying capillary sized blood vessel filled with RBCs and lined by endothelium with no atypia or malignancy.

Clinical discussion: Oral angiokeratoma is a rare disease in patient with Xeroderma pigmentosum that should be early detected and diagnosed to decrease the liability for malignant transformation.

Conclusion: Health case workers should be aware of the nature of the disease, protection of the patient from ultraviolet light as well as Frequent follow up of the patients for a better quality of life.

1. Introduction

Xeroderma pigmentosum (XP) is a hereditary disorder of photosensitivity characterized by the development of different skin lesions in sun-exposed areas of the body, premature skin aging, and neoplasia. Patients with XP show a genetic mutation of abnormal DNA repair [1,2]. Furthermore, XP is associated with macules, angiomas, and telangiectasias [3]. In most cases of XP, malignant transformations as basal cell carcinoma, squamous cell carcinoma, angiosarcoma and fibrosarcoma have also been detected [4]. XP is considered as a risk factor of both melanoma and non -melanoma skin cancers in younger age patients due to the skin susceptibility to ultraviolet radiation [5]. Two-thirds of patients with XP are unaware of their own sickness so it generally advances through an extreme course that requires preventive measures from the patients [5,6].

Searching the literature showed the lacking reports describing oral manifestations of XP. To our knowledge, this is the first case to report intra oral angiokeratoma in a patient with XP. We present a case of 20 years old with XP and angiokeratoma at the right lateral surface of the tongue. The patient reported the absence of a family history of XP.

2. Case history

A 20-year-old male went to National Cancer Institute in Cairo, Egypt in 2008. The patient has a significant medical history of XP for 19 years. Clinical examination of the patient showed pedunculated progressive painful polyps of the scalp as well as multiple nodules in hands that persisted for 14 years. The family history was negative for XP. The lesions were treated with surgical excisions while the histopathological examinations revealed a pyogenic granuloma of the scalp and hand lesions.

In 2011, the patient came back with multiple gradual spreading painful ulcers of the face and scars. After incisional biopsies and the surgical excision, the histopathological examination of the ulcers revealed low-grade squamous cell carcinoma and basal cell carcinoma.

In 2013, the patient developed a 5 * 4 cm ulcer with raised everted ulcer at the buccal mucosa. The ulcer was surgically excised with submental flap and excision with 2 submental lymph nodes (right and left). The histopathological analysis of the cheek ulcer showed poorly differentiated squamous cell carcinoma, while the excised submental lymph nodes were free from any inflammatory changes or signs of dysplasia.

In 2014, physical examination was remarkable for the left cheek, upper labial mucosa, and nasal lesion. The left cheek lesion was

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https://doi.org/10.1016/j.ijscr.2021.106513
Received 14 September 2021; Received in revised form 13 October 2021; Accepted 13 October 2021
Available online 16 October 2021

diagnosed as a non-specific inflammatory lesion while it did not exhibit dysplastic changes. The histopathological examination of the upper labial mucosal lesion revealed a pyogenic granuloma, and it was treated by surgical excision and reconstruction by a rotational flap. The nasal lesion was basal cell carcinoma (morphea type), and it was treated surgically.

In 2015, the patient developed a single sessile, firm, erythematous to violet, painless and shiny papule at the right side of the tongue (Fig. 1). The lesion persisted for 6 weeks. It was surgically excised(Fig. 2) and microscopic analysis of the lesions revealed focally eroded and hyperplastic stratified squamous epithelial covering with underlying capillary sized blood vessel filled with RBCs and lined by endothelium with no atypia or malignancy (Figs. 3). This report is in line with the SCARE criteria [7].

3. Discussion

This report is a unique presentation of intra oral angiokeratoma at the lateral border of the tongue in a case of XP. Ultraviolet damage is considered as the main contributing factor in developing different clinical manifestations in patients with XP [3,8]. Recurrent facial cancers and subsequent surgical treatment of these lesions are considered the most irritating psychological and social implications in XP [2,5].

The detection of benign tumors as angiokeratoma in our case highlights the question of whether XP patients have a higher frequency of developing benign tumors in comparison to the normal population.

Angiokeratomas are vascular malformations of capillaries in the form of single or multiple keratotic papules and/or plaques. It can be mistaken clinically for melanocytic nevus, malignant melanoma, verruca vulgaris, hemangioma, capillary aneurysm, Spitz nevus or focal epithelial hyperplasia. Histopathological examination reveals benign vascular ectasia of the papillary dermis [9]. Intra-oral angiokeratoma was not previously reported in cases of XP.

It is questionable whether the vascular lesions seen in XP patients are reactive lesions due to ultraviolet damage or an increased tendency to develop vascular proliferation as senile angioma-like small red papular lesions and pyogenic granulomas which were detected both in adult and child patients [10].

The management of XP complications, especially orofacial manifestations, represents a considerable challenge to healthcare providers. [1] Wade and Plotnick presented two patients with a history of XP and they developed squamous cell carcinoma of the tongue. Dermal fibroblasts from two black siblings were proven to be hypersensitive to ultraviolet (UV) light by measuring by colony-forming ability [11].

In conclusion, this case is the first reported occurrence of an angiokeratoma of the oral cavity of a patient with XP after different consecutive benign and malignant oral lesions. This case is different from the previous reports of oral abnormalities of XP in which the lesions developed in a non-sun exposed area. Patient education of the nature of the disease, protection of the patient from ultraviolet light as well as Frequent follow up of the patients are essential for a better quality of life in XP patients.

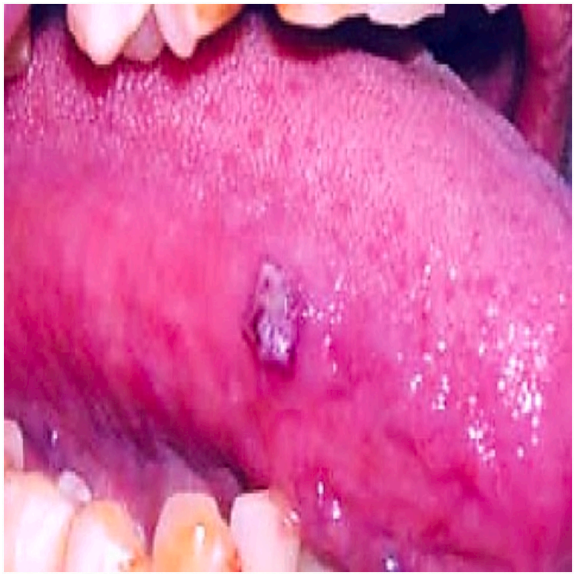

Fig. 1. Single angiokeratoma at the right lateral border of the tongue.

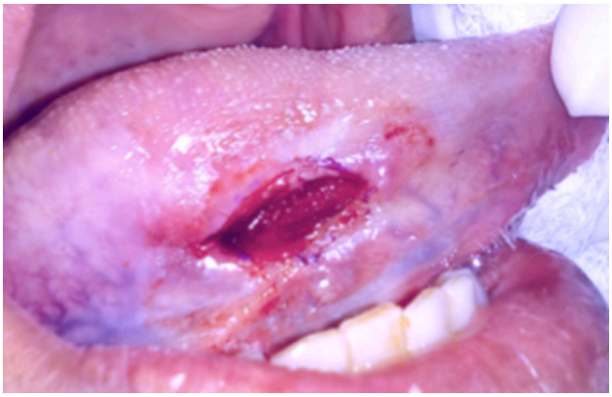

Fig. 2. Surgical excision of the lesion at the lateral border of the tongue.

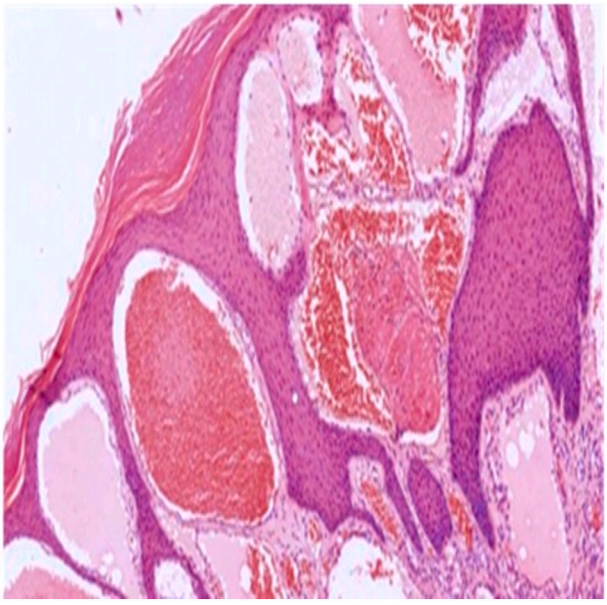

Fig. 3. Photomicrograph of angiokeratoma of the tongue showing hyperplastic stratified squamous epithelial covering with underlying capillary sized blood vessel filled with RBCs and lined by endothelium (Magnification 10×).

Ethical approval

The data presented in the current case report is reviewed and approved by the Ethical Committee at our center.

Patient's consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this

journal on request.

Sources of funding

No sources of funding to declare.

Research registration

Not applicable.

Guarantor

Wafaa Saleh is the Guarantor.

Provenance and peer review

Not commissioned, externally peer-reviewed.

CRediT authorship contribution statement

Wafaa Saleh: concepts, design, literature search, data acquisition, data analysis, manuscript preparation, manuscript editing, and manuscript review

Maha Elansary: concepts, design, data acquisition, manuscript revision

Declaration of competing interest

The authors report no conflict of interest.

Acknowledgement

The authors deny any conflicts of interest.

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