Epidermodysplasia Verruciformis Associated with Cutaneous Malignancy: A Rare inherited Genodermatosis

Dr. Anne Cinthia Ashwin – M.D. (D.V.L.), Dip. Aesthetic Medicine (AAAM)

Introduction

Epidermodysplasia Verruciformis (EDV) is an inherited disorder associated with defects in cell mediated immunity, characterized by multiple plane warts, pityriasis versicolor-like lesions and a tendency to develop skin malignancies.

Here, we report a familial case of Epidermodysplasia Verruciformis in a mother and daughter, who developed squamous cell carcinoma in sun-exposed areas.

Case Report

A 29-year-old female presented with multiple asymptomatic elevated skin lesions over the face, arms, forearms, and legs persisting for the past 26 years. She has been on oral and topical treatments ever since. The patient's mother also had similar complaints since her young age and developed cancerous lesions over her face.

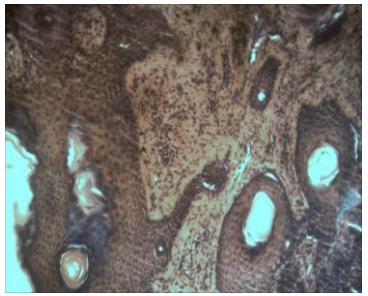
On examination, multiple hyper pigmented plane to warty, papule to plaques, with hypo pigmented macules were observed over sun-exposed areas. A 2x3 cm ulcerated plaque was present on the forehead, which bleed on touch, with everted edges and a crust-covered base.

Diagnosis

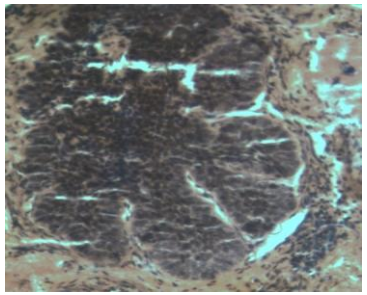
Skin biopsy taken from the warty papule of right forearm.

Histopathology features

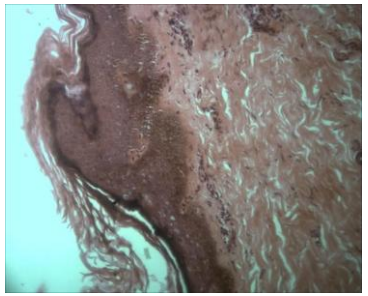
A: The tumor shows buds and irregular proliferations of tumor tissue attached to the undersurface of the epidermis. Epithelium and stroma form reminiscent of follicular germs.
B: The islands of tumor cells show peripheral palisading pattern, as well as mitotic and apoptotic figures.
C: The epidermis is hyperkeratotic and acanthotic. A few dyskeratotic cells are seen in the lower part of the epidermis.
D: Large affected keratinocytes are swollen and irregularly shaped and shows abundant blue-gray cytoplasm. Keratohyalin granules are prominent in the granular layer.



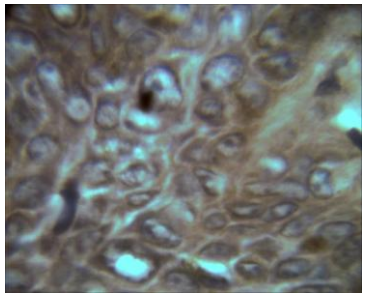
A. H&E, 40x



B. H&E, 40x



C. H&E, 40x



D. H&E, 40x

Discussion

Epidermodysplasia Verruciformis (EDV) is an inherited autosomal recessive gene disorder. EDV is rarely seen in immunocompromised adults, though it is more commonly observed during childhood in individuals with a family history of the disease. The disease is mostly linked to mutations in the EVER1 and EVER2 genes. There are over 20 HPV types characteristic of EDV, of which types 5, 8 and 47 are most associated with malignant EDV lesions. Up to 90% of EDV patients between the age group of 20 – 40 years, develop nonmelanoma skin cancer in sun exposed areas, mainly squamous cell carcinoma.

Conclusion

Early recognition of EDV, which is a rare cutaneous disorder, is of paramount importance for physicians to counsel patients about its potential to develop into skin malignancies. This can enable early diagnosis, which could subsequently lead to the management of the disease, resulting in an improved patient outcomes.

Reference

- da Cruz Silva, L.L., de Oliveira, W.R.P. & Sotto, M.N. Epidermodysplasia verruciformis: revision of a model of carcinogenic disease. Surg Exp Pathol 2, 20 (2019).
- RobertJ.Ragotte,StuartE.Turvey,EVER1andEVER2MutationsinEpidermodysplasiaVerruciformis,EncyclopediaofMedicalImmunology,10.1007/978-1-4614-8678-7,(303-308),(2020).