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EPIDERMODYSPLASIA VERRUCIFORMIS ASSOCIATED WITH OCULAR SURFACE SQUAMOUS NEOPLASIA.

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ABSTRACT **INTRODUCTION**: Epidermodysplasia verruciformis is a rare genodermatosis characterized by a unique susceptibility to cutaneous infection by a group of phylogenetically related human papilloma viruses (HPVs). These patients show a defect in cell-mediated immunity specific toward the causative HPVs that lead to lifelong disease. The defect is usually inherited as autosomal recessive trait and presents clinically with plane warts, pityriasis versicolor-like lesions and reddish verrucous plaques. Dysplastic and malignant changes in the form of actinic keratoses, Bowen's disease and squamous cell carcinoma (SCC) are common but metastasis occurs rarely. A totally effective treatment against EV is as yet highly desirable

MATERIALS AND METHODS:

CASE REPORT-

A 28 year old man, born of consanguineous marriage ,a known case of epilepsy, presented with multiple extensive erythematous , hyperkeratotic and verrucous plaques over trunk, arms and legs , hypopigmented scaly macules over the face and dystrophic nails for the past 10 years.No similar Family history . Sexually transmitted infections were excluded by Rapid plasma reagent test and ELISA (HIV) test, also patient denied high risk behavior.

We thought of verrucous xeroderma pigmentosum. Hence skin biopsy was done. Histopathology findings were consistent with epidermodysplasia verruciformis . Patient also had a fleshy growth in the right eye which increased in size , resulting in decreased vision in the past one year. Impression cytology showed features consistent with ocular surface squamous neoplasia in the right eye by the ophthalmologist.

DISCUSSION : The association of OSSN and EV has been reported only once in the literature. OSSN in younger age groups should arose suspicion of systemic predisposing diseases. A through dermatological and opthalmological evaluation and follow up will help us rule out such association.

KEYWORDS : Epidermodysplasia verruciformis , papilliform type, ocular surface neoplasia, Human papilloma virus.

BACKGROUND.

Epidermodysplasia verruciformis (EV) is a rare hereditary disease. The exact prevalence of EV is unknown but more than 200 cases have been reported in the literature so far. In early childhood, scaly macules appear on the trunk, shoulders, neck, arms and face. They may, however, be more widely distributed. The lesions are brown, red or white and in darkly pigmented skin they may appear black. The appearance and distribution of these lesions has been likened to pityriasis versicolour. Hyperpigmented verrucous lesions are seen in papilliform type of EDV [1].Seborrheic keratosis, actinic keratosis and squamous cell carcinoma arise in sun-exposed areas, most commonly on the forehead. Malignant conversion of the benign lesions occurs in more than half of patients who have had the disease for 20 years or more and can occur on any sun-exposed area.[2,3.]

Ocular surface squamous neoplasia (OSSN) encompasses the entire spectrum of dysplastic and carcinomatous lesions of the ocular surface [1].OSSN may represent abnormal maturation of corneal and conjunctival epithelium as a result of a combination of factors such as ultraviolet B irradiation and human papilloma virus. Other reported risk factors include exposure to petroleum products, heavy cigarette smoking, chemicals such as trifluridine, arsenicals and beryllium, and ocular surface injury, vitamin A deficiency, light pigmentation of the hair and eyes, and HIV infection. [2,3.]

CASE PRESENTATION

A 28 year old man, a painter by occupation presented with multiple extensive erythematous , hyperkeratotic and verrucous plaques ,discrete verrucous papules over trunk, arms and legs. Multiple hypopigmented erythematous scaly macules over the face, neck, trunk, arms , leg were present . Confluent hyperkeratotic plaques over the arms (below elbows) and legs (below knees) , palms and soles were present . The lesions started when he was 8 years old , since then it gradually increased in size and number , progressing to involve the entire body surface. Scalp and hair were sparred. Dystrophic nails were present. Few verrucous papules seen over the lips, aphthous ulcers and angular chelitis was present. No family members had a similar affliction. The patient was born of consanguineous marriage . History of epilepsy till 5 years of age present. Patient denied high risk behavior. No systemic complaints present.We thought of verrucous xeroderma pigmentosum. Hence skin biopsy was done. Histopathology displayed epidermal thickening with swollen cells in the upper epidermis , nests of large cells with perineuclear halo and greyish blue cytoplasm present, basket weave pattern of stratum corneum and few dysplastic cells were seen in the epidermis. Findings were consistent with epidermodysplasia verruciformis . Patient also had a fleshy growth in the right eye which increased in size , resulting in decreased vision in the past one year. On slit-lamp biomicroscopy, the right eye showed an ill-defined, elevated, gelatinous nodular mass (6×4 mm) at temporal limbus (7–10 o'clock hours). Another similar mass (2×2 mm) was present at the nasal limbus (5 o'clock hours). Feeder vessels were also visible. No family members had a similar affliction. Conjunctivalisation of the cornea was appreciable along all the masses. Gonioscopy did not show any abnormality . Lens and retinal examination of both eyes was normal. No lymphadenopathy was present.

Investigations

Impression cytology from the mass in the right eye showed features of mild squamous epithelial cell dysplasia along with neutrophilic infiltration. Ultrasound biomicroscopy examination of the eyes did not show any deep infiltration of the mass into the sclera. A diagnosis of EV was made based on histopathology of the skin biopsy . PCR analysis from the skin lesions was positive for β-1 human papillomavirus (HPV-5). An ELISA test for HIV was negative. RPR was negative.

DISCUSSION

EV is a rare, inherited disorder that predisposes patients to widespread HPV infection and cutaneous squamous cell carcinomas[4,5.] The most common inheritance pattern is autosomal recessive, though rarely, autosomal dominant and sporadic patterns may be seen. The disease is characterised by chronic infection with β-HPV. Widespread skin eruptions of flat-to-papillomatous, wart-like lesions and reddish-brown pigmented plaques on the trunk, hands, upper and lower extremities and face, are typical.

More than 30 HPVs have been identified in EV lesions. HPV-5 and HPV-8 have been isolated in more than 90% of EV–associated squamous cell carcinomas. Treatment of EV is limited and aimed mainly at preventing the progression of benign lesions into malignant ones. No therapy for EV is definitive. Treatment of EV includes preventive measures, the most important of which is strict sun avoidance and protection, beginning as soon as the diagnosis is made.

The association of OSSN and EV has been reported only once in the literature. Partridge and Pariser[6] reported a case of ocular and cutaneous squamous cell carcinoma in an African-American man with EV resulting in blindness and death.

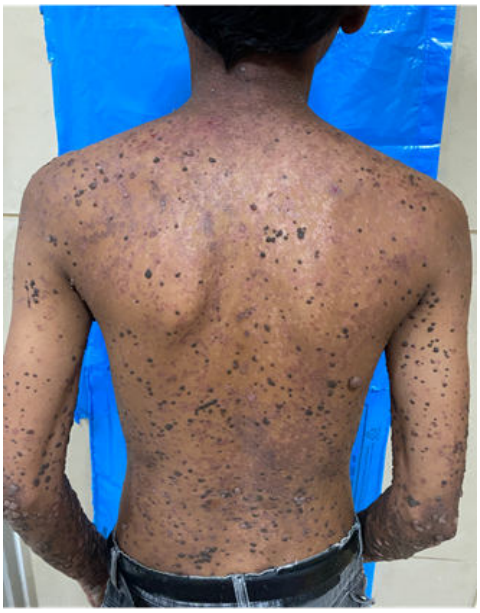

FIGURE 1 : verrucous plaques and hypopigmented macules in the trunk

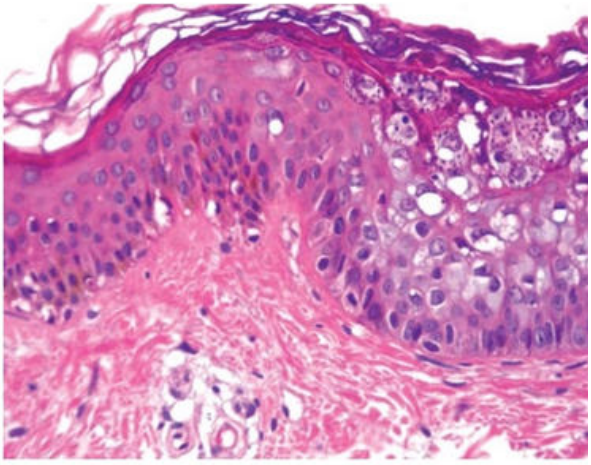

Figure 2 : Histopathology Picture Of Edv- Basket Weave Hyperkeratosis , Dysplastic Cells In Epidermis.

FIGURE 3 and 4 : confluent verrucous plaques in palms. Dystrophic nails seen.

FIGURE 5 : fleshy growth in the right eye

Patients with EV are prone to having multiple malignant lesions and their prognosis is worse as compared to older individuals without any systemic disease.

This case report highlights the fact that occurrence of OSSN in younger age groups (<30 years) should arouse suspicion of any systemic predisposing disease such as xeroderma pigmentosum and HIV infection. EV, though rare, is associated with OSSN. A thorough dermatological evaluation and follow-up will help rule out such association.

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