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Case Report:

Ocular Surface Squamous Neoplasia in Xeroderma Pigmentosum

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Abstract: Xeroderma pigmentosum (XP) is a rare genetic disorder associated with multiple oculocutaneous and neurological manifestations. It occurs due to deficiency of the enzymes responsible for repairing ultraviolet radiation-induced DNA damage. Persistence of un-repaired DNA results in somatic mutations, leading to neoplasia of the skin and ocular surface. As this condition is rare, only isolated case reports of XP with ocular surface squamous neoplasia (OSSN) are found in literature.
Key Words: Ocular surface squamous neoplasia; Xeroderma pigmentosum

Case Report:
Six years old male child diagnosed with Xeroderma Pigmentosum was referred to ophthalmology from dermatology department. He presented with mass in the right eye since 3 months, gradually progressing in size and painless. He had bilateral photophobia. There was a similar history of pigmented skin lesion in the sibling. On examination, visual acuity was perception of light and 6/60 in the right and left eye respectively.
Ocular motility and left eye fundoscopy was normal. Right eye showed fleshy pink irregular pedunculated mass lesion of 3 cm arising from superior limbus covering the cornea.[Figure 1] Left eye showed hazy cornea and hyperpigmented 4mm lesion at 7'O clock position, conjunctival naevi at 3'O clock and 5 o clock hours close to limbus
Right eye mass was excised and sent for histopathology examination. Histopathogy report revealed well differentiated squamous cell carcinoma with extensive hyperkeratosis, acanthosis and stromal invasion[Figure 2]. Post-operative photogragh of the patient after excision of mass lesion is shown in Figure 3.

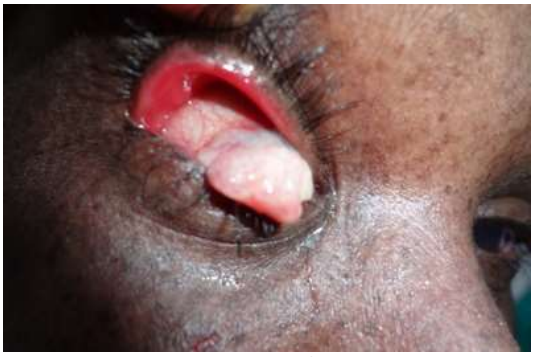

Figure 1: Fleshy mass arising from superior limbus covering the cornea of the right eye

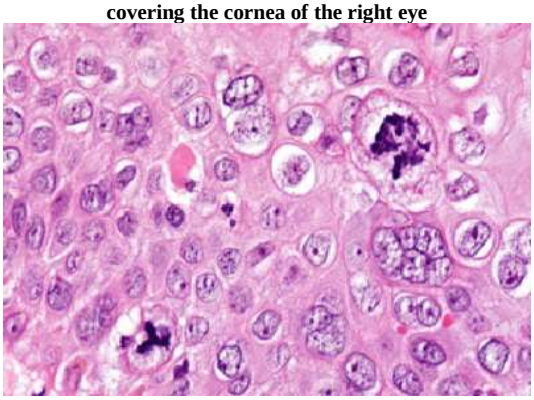

Figure 2: Histopathology showing well differentiated squamous cell carcinoma

Figure 3: Post operative photogragh after excision of mass lesion

Discussion:

Ocular surface squamous neoplasia (OSSN) includes a spectrum of diseases which range from mild dysplasia to conjunctival and corneal intraepithelial neoplasia (CIN) and invasive squamous cell carcinoma (SCC). Pre-invasive OSSN lesions are classified as mild, moderate, or severe, depending on the degree of involvement of the dysplastic epithelium. Mild dysplasia, or CIN grade I, is defined as dysplasia confined to the lower third of the conjunctival epithelial thickness. Moderate dysplasia (CIN grade II) extends into the middle third, and severe dysplasia (CIN III) to the upper third of the conjunctival epithelium. Full-thickness dysplasia of the epithelium is also referred to as carcinoma-in-situ. The pathological basis for a diagnosis of pre-invasive OSSN is based on the identification of epithelial disarray and abnormalities in maturation. In general, these conditions show an abrupt demarcation between neoplastic cells and the uninvolved benign epithelium. Invasive OSSN or squamous cell carcinoma shows nests of infiltrating cells that have penetrated the epithelial basement membrane and spread into the conjunctival stroma.

Xeroderma pigmentosum is inherited as an autosomal recessive disorder with a prevalence rate of 1:250,000. It occurs due to deficiency of the enzymes responsible for repairing ultraviolet radiation-induced DNA damage.[1] It is characterized by multiple pigmented spots, called 'freckles' and larger atrophic lesions, and a glossy white thinning of the skin.[2] Conjunctival and corneal epithelial malignancies are seen more commonly in elderly and male patients, but may develop at a younger age, especially in association with xeroderma pigmentosum or immunodeficiency. OSSN predominantly occurs in older males with an average age of 56 years.[3] This patient presented with OSSN at 6 years of age. Patients with XP are unable to repair the DNA that is damaged by ultraviolet rays. This can lead to somatic mutations and development of cancerous cells. This inherent defect accounts for the increased susceptibility to OSSN and the younger age at presentation.[4] Some reports have noted a younger age in intraepithelial cases compared with invasive squamous cell carcinoma.[5] OSSN most commonly occurs at the limbus, as it is a transition zone and is prone to development of dysplasia.[3] Conjunctival squamous cell neoplasms can cause significant ocular morbidity. Early diagnosis and intervention can prevent extensive visual morbidity. Simple excision of conjunctival intraepithelial or invasive neoplasia has been reported to be associated with a 24–50% recurrence rate.[3-6] Excision with intraoperative control of the surgical margins and adjunctive cryotherapy has been reported to reduce recurrence rates to 12%.[7-8] A majority of the patients of XP with OSSN develop recurrence of the tumor despite a meticulous tumor excision and adjunctive cryotherapy. Fresh lesions may also be encountered during the follow-up examination of these patients. The increased risk of recurrence of OSSN in patients of XP warrants a regular, meticulous follow-up with an ophthalmologist.

Conclusion: OSSN occurs predominantly in elderly, but in patients of Xerdoderma Pigmentosum it tends to occur at a younger age (6-22 years), with a predilection for the limbal area. As XP patients have potential blinding complications, an ophthalmologist must be involved from the beginning in the care of these patients. Awareness and prompt management with close follow up for ocular and cutaneous malignancy is warranted in these patients.

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