Squamous Cell Carcinoma Face with Gigantic Neck Metastasis in a Child of Xeroderma Pigmentosum: A Rare Case Report

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ABSTRACT
Background: Xeroderma pigmentosum is a rare autosomal-recessive (AR) disorder due to defective DNA repair mechanism that appears in early childhood. It is a known premalignant condition. Squamous cell carcinoma (SCC) is not uncommon in patients with xeroderma pigmentosum and mostly involves the face, head, neck, and scalp. However, SCC of the face may exhibit an aggressive course. Here, we present a case of xeroderma pigmentosum with SCC of the face with gigantic neck metastasis in a ten years old child. In xeroderma pigmentosum patients, squamous cell carcinoma of the face can present early and tends to be unusually aggressive. So proper education to the patient and their parents about ultra-violet light protection, especially in sunny areas, and early recognition of any suspicious lesion could be life-saving.

Keywords: Aggressive, Squamous cell carcinoma, Face, Neck, Xeroderma pigmentosum

INTRODUCTION
Xeroderma pigmentosum (XP) is a rare autosomal recessive genetic disorder in which nucleotide excision repair (NER) enzymes are mutated, leading to a reduction in or elimination of NER in UV damaged DNA, and a few patients with defects in “postreplication” or “daughter-strand repair” are known as “XP variants”. XP is a well documented pre-cancerous state for skin cancers viz squamous cell carcinoma (SCC), basal cell carcinoma (BCC) and melanomas. These malignant tumors mostly involve the face, head, neck, and scalp. Early symptoms are observed in the sun-exposed parts of the skin and eyes and include poikiloderma, skin atrophy, and telangiectasias. Oral cancers, leukoplakia, and cheilitis are common oral manifestations. Education of the patient with XP and his family about strict avoidance of sunlight, especially in sunny areas and early presentation for any suspicious lesion in sun exposed area will improve the results.

Case report
A ten-year-old boy, product of consanguineous marriage, known case of XP, presented to our department with ulcerating masses in the face and right hand. The ulcers were round in shape, measured about 4 × 6 cm², with punched out edges. There was discharge but no bleeding. At presentation, he was irritable and photophobic. There were diffuse hyperpigmented lesions over the sun exposed areas like face, trunk, and extensor surface of the upper extremities. The skin was unnaturally dry and rough all over these areas. Hyperpigmented hyperkeratotic changes were observed over facial lesions. There was a congestion of conjunctivae of both eyes. The scalp was covered with multiple scaly plaques.
Neurological examination was normal. He has a family history XP in two siblings with mild disease. Biopsy showed moderately differentiated squamous cell carcinoma [figure 1]. The patient presented with a late diagnosis of SCC that has been progressing for 3 years. Wide local excision (WLE) of the lesions with 1 cm safety margin circumferentially was done followed by split thickness skin grafting (STSG) [figure 2]. Histopathology examination revealed squamous cell carcinoma (SCC) with free circumferential and deep margins. One year later the patient presented with recurrence in the form of multiple small ulcerations on face with gigantic neck swelling. Fine needle aspiration of the neck swelling revealed deposits of SCC. The neck swelling was increasing in size very rapidly and involved whole of the neck, right lower half of the face, destroyed the nose. Due to rapid increase in size, the overlying skin got necrosed followed by ulceration and bleeding [figure 3]. Patient was referred to hospital tumor board for further management. A written informed consent was obtained from the parents for publication of this case report and any supplementary image.

Discussion

XP is a rare autosomal-recessive disorder that appears in early childhood. It was first described by Hebra and Kaposi in 1874. In the United States, the estimated prevalence of XP is at 1:1,000,000. XP can be diagnosed when a child presents with multiple pigmented lesions, marked photosensitivity, and xerosis. There is a 10,000-fold increased risk of non-melanoma skin cancer under 20 years old. Many risk factors are known to exacerbate the cutaneous features resulting in numerous pigmentation changes, various skin cancers, and early death. These risk factors include sunny weather, outdoors living, fair skin, smoking, poor availability of diagnostic facilities, delayed diagnosis, and poor protection from sunlight.

In patients with XP, the mean age for skin cancer is 8 years compared to 60 years in the healthy individuals. Actinic damage occurs between 1 and 2 years. The most common types of cancer found in XP patients are squamous cell carcinoma and basal cell carcinoma, mostly involving the face, head, and neck. SCC in the face tends to have an aggressive course. Cutaneous neoplasm in XP patients is difficult to be prevented. However, early protection from ultra-violet (UV) radiation may play a role in prevention of skin malignancy. Individuals with XP should strictly avoid sun exposure; use appropriate clothing, sun blocking agents, and protective glasses. In addition, early diagnosis of any skin lesion is of utmost importance. Follow up on a regular basis is critical to detect and excise pre-cancerous lesions and malignant tumors at an early stage. All families at risk should be offered genetic counseling.

Conclusion:

Clinicians should be aware of the possibility of early onset of SCC when patients with XP present with any skin lesion, especially at the exposed areas like face. SCC of the face can be unusually aggressive. Proper education to the parents about the importance of strict UV light protection, especially in sunny areas, is warranted. Early recognition of any suspicious lesion may be life-saving.

References


Figure 1: showing histopathology slide with prominent keratinization with pearl-like structures-keratin pearls suggestive of invasive SCC

Figure 2: showing hyperpigmented and hyperkeratotic lesions over the hand with well settled skin graft over the dorsum
Figure 3: showing gigantic metastatic swelling in the neck extending to face with the overlying skin necrosed and ulcerated. Nose of the patient is eroded by the tumor