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Case Note

Xeroderma pigmentosa – A disfiguring disease

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Abstract
Xeroderma pigmentosa is a rare inherited autosomal recessive disease with the inability to repair DNA damage caused by UV light. Recognized in the late 1800 by Maritz Kaposi it has been reported world wide and in al1 races with an over prevalence of 1-4 per million population. Kunwar et all[1,2]. Those affected are extremely sensitive to the UV portion of the light and have a 2000 fold increased risk of skin cancer in the sun exposed skin. Basal cell carcinoma is the most commonly associated carcinoma followed by Squamous cell carcinoma and Melanoma. The pigmentation on the face and the rest of the body can be horribly disfiguring. The recurring cancer occurring on the face and repeated surgical treatment for the ulcerations have important social and psychological implications not encountered with other cancers[3]. We report two cases of BCC and melanoma. The first case is of BCC of the face in a teenaged girl coexisting with xeroderma pigmentosa. The second case presented with melanoma of the scalp in a 10years old female child. The details of these cases are presented and the management.

Key Words: Xeroderma pigmentosa, Autosomal recessive, Basal cell carcinoma, Squamous cell carcinoma, Melanoma.

Inherited as an autosomal recessive trait Xeroderma pigmentosa is genetically heterogeneous with at least 7 different variants[4]. Those affected are extremely sensitive to the UV portion of light. They rapidly develop skin atrophy, splotchy pigmentation, telangiectasia and skin cancer.

Case Report: 1
A 17years of age female patient who had already been diagnosed as a case of Xeroderma pigmentosa two years back had presented with history of skin pigmentation over the face, neck and the limbs for 10 years, freckles over the face and photophobia. The trunk had been spared. Ulceration 2 by 1 inch over the nasal bridge was present for two years. She gave history of similar ulcerations over the cheeks two years back which had been excised and grafting. No recurrence has occurred on these grafted areas. Biopsy report of the excised portion had revealed BCC. She also received 6-7 weeks of radiotherapy for the same. Her younger brother was unaffected with the disease though she hasn't seen him for the past two years.

Local examination revealed ulcerations extending over whole of the nasal bridge and the root of the nose also involving the medial canthus with crusting and haemorrhage. She had small ulcerations on the forehead .She had two areas of previous skin graft on either cheeks. Small pustular and papular lesions over the body sparing the trunk were also seen. No significant cervical lymphadenopathy was present. Systemic examinations including neurological function were normal.
All investigations with serum biochemistry were within normal limit.

Management:
She was managed with surgical excision of the basal cell carcinoma of the nose. Wide surgical excision of the carcinoma with the trimming of the medial nasal septum and some excision of the right alae cartilage which appeared to be involved in the ulcer. It was decided to cover the nasal defect with midline forehead flap based on the right supraorbital and supratrochlear vessels. As the skin was insufficient on the left side a small left cheek skin was added by doing a VY plasty. The forehead was having many small ulcers so the whole forehead skin was excised and a thick skin graft from the upper arm was applied.

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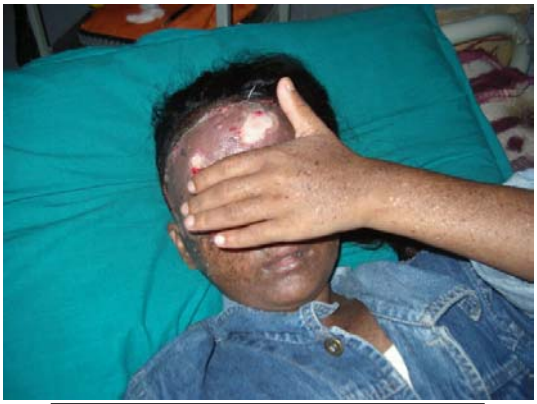
Fig. 1

After excision ulcer nose, forehead flap for cover and excision forehead ulcers and SSG. Small areas of skin graft had necrosed. Bare bone drilled to promote granulation and grafted.



Patient suffering photophobia

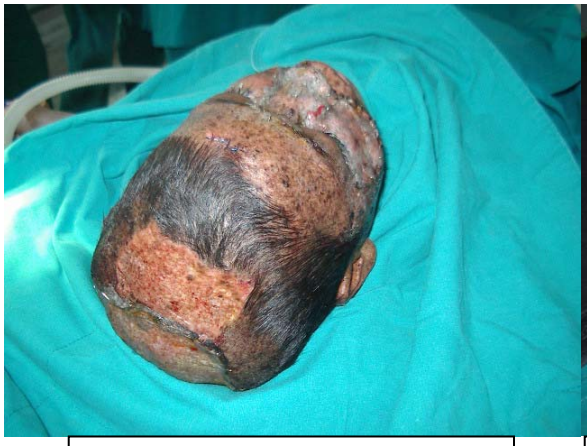
Case Report 2

A 10 years old female child presented with the chief complaints of pigmentations of the skin all over the body since 8 years. According to the patients father it had started from the scalp when she was 2yrs of age. Then it gradually progressed to all the parts of the body. The lesions were initially scaly which eventually peeled off giving rise to ulcers .The pigmentation increased following exposure to the sun. Swelling over the scalp region occurred eight months back and another similar swelling over the left lower eyelid five months back. Excision of the mass with skin grafting done was eight months back. Histopathological report revealed Malignant Melanoma.

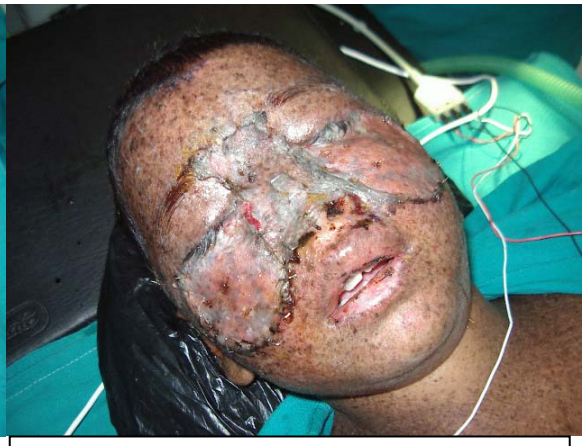
On examination, General condition of the patient was fair. Systemic examinations were normal. Neurological status-intact.

Local examination- Diffuse pigmentation all over the body with pappilomatous growth ranging from approx 3 x 5 cm in size on the scalp with ulcerations and foul smelling purulent discharge. A growth measuring approx 1 x 0.5 cm over the right bridge of the nose and another growth measuring 1 x 1 cm over the left lower eye lid were present.

Fig. 2



Excision Scalp lesion and SSG done



Excision lesion nose and both cheeks and SSG

Discussion

Inherited as an Autosomal Recessive Trait XP is genetically heterogeneous with at least seven different variants .It was recognized in the late 1800 by Maritz Kaposi. Those affected are extremely sensitive to the UV portion of the sunlight and have a 2000 fold increase in the development of skin cancers in the sun exposed portion of the skin which tend to recur.

The discovery of the defect in the repair of the UV light induced damage to the genetic material was made in the 1960s when a radiobiologist J.Cleaver investigated the Ultraviolet radio sensitivity *in vitro*. He had heard about the San Franciaco "moon people" so called because they knew that they had to avoid the sunlight in order to prevent recurrent skin cancer. This discovery brought out the clinical relevance to years of fundamental research in microbes and the interaction with radiation resulting in mutagenesis and carcinogenesis.[5]

The most important biochemical effect of the radiation is the formation of pyrimidine dimmers in the DNA. This is normally repaired by the Nucleotide excision repair enzymes. (NER)This process requires the product of at least 20 genes. The defect in the gene leads to the inability to repair the damaged

DNA and thus there is the occurrence of sunburn that does not heal following minimal sun exposure. Other features include blistering, cutaneous telangiectasia,

increasing irregular pigmentation, crusting of the skin, scaling of the skin, photophobia and skin cancers. Malignant skin lesions are often present before the child is 5 years old. Neurological damage (20%) also occurs like deafness, developmental delay, spasticity and depression and psychological disorder

Diagnosis is made by Family history of XP. Other than that examinations may reveal clouding of the cornea, keratitis, lid tumours and blepharitis. There are a number of tests that help in diagnosing the disorder before and after the birth of the child such as amniocentesis and villous sampling and of course, skin biopsy.

Management

There is no specific treatment for XP and management relies on preventing the damage where possible and dealing with the damaged and aberrant tissues at an early age.

1. Total protection from sunlight. Also, prevention of light coming through the window glass and light from the fluorescent bulb which can also be dangerous. Restrict outdoor activities to night time Protective clothing must cover the skin
2. High protection (SPF 30 or greater) sunscreen and UV protected glasses should be worn.
3. Regular dermatology, ophthalmology and neurological examinations must be done
4. Early removal of skin lesions when detected.
5. Genetic counselling should be arranged for parents of an affected child.

6. Psychological counselling.
7. New options that are still under research include T4 endonuclease delivered topically used with sunscreen agents .It decreases the incidence of BCC and actinokeratosis. Chemoprevention with Green tea and soy are still under research. Retinoids may suppress the development of malignancies.
8. Surgical management consist of Electrodissection, Excision, Cryosurgery Moh's surgery and wide local excision. Radiotherapy may be given. Depending on the severity of the location of the cancer reconstruction option can range from simple closure with or without grafting and flaps.

Conclusion

Xeroderma Pigmentosa is a horribly disfiguring disease that needs the expertise of dermatologists, plastic surgeons, rehabilitation experts and sociologist. Many of these patients are socially ostracized because of this disfigurement. They have to be viewed with compassion and patience.

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