

Xeroderma pigmentosum with Malignant Ulcer Associated with Plenty of Live Maggots—A Rare Case Report

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Abstract: *Xeroderma pigmentosum (xp) means dry pigmented skin . It is a rare genetic disorder with extreme sun –sensitivity to UV light . UV light damages genetic material (DNA) in cells and disrupts normal cell function. This genetic condition is associated with increased risk of childhood cancer. Here is the case report of XP with Squamous cell carcinoma of skin with rare, strange association with plenty of live maggots.*

Keywords: Xeroderma pigmentosum, sun-sensitivity, Ultraviolet light, Genetic material, Squamous cell Carcinoma, maggots.

1. Introduction & Literature Review

The condition is more common in Japan , North Africa and middle east. XP , a familial cancer syndrome, a rare autosomal recessive genetic disorder is associated with extreme photosensitivity and defect in the damaged DNA repair abnormality

Estimated incidences vary from 1 in 20, 000 in Japan to 1 in 250, 000 in the USA, and approximately 2.3 per million live births in Western Europe.¹ It effects both men & women, has increased familial or genetic susceptibility to malignancies- basal cell carcinoma, squamous cell carcinoma and melanomas, inherited by autosomal recessive pattern, the parents carry each one copy of mutated gene but they do not show any signs and symptoms of this condition. The disease is a serious mutilating disorder and the life span of the affected patient is often brief.²

The skin lesions manifest during infancy or early childhood over sun-exposed areas such as face, neck, arms, hands and scalp⁴.

They include erythema, bullae, scaling . crusting, keratosis , telangiectasia, basal and squamous cell carcinoma and malignant melanoma.

Ocular manifestations include photophobia lacrimation, blepharitis symblepharon, keratitis, corneal opacities, tumours of the lids, and possible eventual blindness.³

NEUROLOGICAL ABNORMALITIES such as mental deterioration and neural deafness may develop in 20% of the patients.

The most important part of the treatment is avoiding exposure to sunlight.

2. Case Report:

A 5 yr old female child with non-consanguineous parentage with normal developmental milestones and with younger female sibling with similar lesions was admitted in paediatric department of Medical college/teaching Hospital, Guntur with chief complaints of generalised skin rash and swelling of 7X7 cm size over the right side of the cheek and

temporal region which ruptured with foul smelling, bloody pus discharge which was associated with pain and itching. There was continuous high grade fever with chills and rigors. Patient had photophobia and congestion of eyes .There was a foul smelling discharge from both the ears.

The child was apparently well up to the age of 5 months after birth. Then the skin lesions appeared with small blackish macules over the scalp and face. Later they became papules and spread over to abdomen, upper limbs, lower limbs, and back. They are associated with severe itching . Photophobia to sunlight and fluorescent tube light developed at the same period.

The skin and eye symptoms gradually progressed since then. *Patient also had foul smelling ear discharge with shedding thick casts from both ears since then.*

3. Examination

The skin lesions are present all over the body , more over the sun exposed parts including the scalp.(fig-1 & fig-2).



Figure 1



Figure 2

Eye examination revealed swollen eye lids with conjunctival congestion, chemosis with clear cornea and with marked photophobia. Examination of ears showed intact normal tympanic membranes with otitis externa. Other systemic examination was normal.

Swelling with Abscess over right temporal region and cheek measured 7X7 cm in size, on removal of the upper layer multiple live maggots with foul smelling bloody-pus discharge were seen.(fig-3).



Figure 3: XP, malignant ulcer with maggots



Figure 4: ulcer healed well after chemotherapy.

Multiple pustules with itching were present over the scalp. Photophobia with eversion of the lower lids was marked.

4. Investigation S And Management:

Blood picture revealed hypochromic microcytic anemia with neutrophilia. HIV & HBSAg were non-reactive. Blood culture & sensitivity revealed *Pseudomonas aeruginosa* sensitive to ceftaxone, azithromycin and amikacin. The ulcer was filled completely with plenty of live maggots. The maggots live on dead decaying organic matter. As it was difficult to remove them mechanically we used chloroform for their removal. The ulcer was cleaned well and dressed. Debridement of the wound was done with daily dressing.

Maggots reappeared again on 2nd and 3rd day, emerging out from underlying soft tissues, probably from narrow burrows beneath the ulcer. Repeated ulcer cleaning was done 3,4 times and cleared of the maggots.

Wedge Biopsy of the ulcer specimen subjected to histopathological examination and report revealed a well differentiated squamous cell carcinoma.

5. Treatment

Surgery –depends on whether tumor is resectable and on the actual extent of resection. Because of the invasive nature of maggots surgery is deferred.

Chemotherapy –most widely used; followed in order of use, by surgery, radiation therapy, and biological agent therapy. Radiation therapy is used sparingly in children because they are more vulnerable than adults to its late adverse effects, and for those tumors with incomplete response to chemotherapy.

To avoid future recurrence of malignancies at various sites like –skin, mucous membranes and eyes, Chemotherapy was planned with IV cyclophosphamide 200 mg. once in a week. This patient received 4 cycles of the drug and the ulcer healed well.(fig-4)

IV ceftriaxone for control of infection was used.

Iron folic acid and multivitamin supplementations, methyl cellulose eye drops were given.

Photo protection with sun glasses, broad spectrum sun screen lotions and creams were advised. Full coverage of the scalp and body with clothing advised.

Advised to avoid indoor fluorescent tube light source of UV light.

6. Follow Up

On follow up; symptoms improved and ulcer healed well. But there is recurrence of same skin lesions without ulcer on periodic follow up visits over 3-4 months.

7. Discussion

The most common types of cancer found in XP patients are basal cell cancer and Squamous cell cancer with most tumours found on the face, head or neck .2

There are very few reports of other types of cutaneous neoplasms including angioma or fibrosarcoma. Additionally, there is a 10- to 20-fold increase in the frequency of occurrence of lung, uterine and CNS neoplasms in patients with XP. Anterior tongue neoplasms have been reported and are presumably due to direct exposure to the sun.3

The exact genetic defects are still not fully understood in all forms of XP, but defective repair of UV induced DNA damage is found in most individuals. Defective repair replication was later reported with dermal fibroblasts, lymphocytes and conjunctival cells.5

Myiasis is that caused by the necrobiophagous flies (feed on dead tissues). This is the more common type and attacks patients with necrotic cavity lesions. They are photophobic, they hide in deep tissues by making burrows⁶. They obtain nourishment from the tissues; and in this stage, there will be tissue inflammation ensuing discomfort. The larvae are

voracious and destroy integral tissues and may cause serious hemorrhage which can even prove to be life threatening 7.

Where as is in case of MDT Wound debridement, originally thought to be a mechanical effect of the maggots,⁸ has been shown to be due to three proteolytic enzyme classes that were identified in the maggot excretions.⁹ Maggot excretions have an inhibitory effect on both Gram-positive and Gram-negative bacteria including methicillin-resistant *Staphylococcus aureus*, methicillin-sensitive *S aureus*, *Escherichia coli*, and *Pseudomonas aeruginosa*. The ammonia excreted by maggots is believed to alter the pH of the wound, which inhibits bacterial growth.¹⁰

In this cases all features of XP were seen , the disease is progressive after a period of remission with treatment . There was recurrence of a skin, eye symptoms. As these patients have functional defects of immunity system,this patient is predisposed to bacterial infections with pseudomonas and streptococci of blood and discharges.

Maggots were seen in multiples, they were invasive in the wound, cleared of the ulcer with much difficulty with chloroform and turpentine oil applications, these applications stimulate the maggots to come out from the deep burrows. they thrive on dead and decaying organic matter in tissues. As the invasive nature of maggots and ulcer, chemotherapy is preferred to wide surgical excision. Debridement of the wound was done.

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