Original Article

Xeroderma Pigmentosum

Ocular Manisfestations in Xeroderma Pigmentosum; A Series of 20 Patients

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ABSTRACT

Objective: To assess the various ocular manifestations in xeroderma pigmentosum seen at Eye and Dermatology Department of Chandka Medical College Hospital Larkana.

Study Design: Prospective study

Place and Duration of Study: This study was carried out at two Departments, i.e. at Departments of Dermatology and Ophthalmology of Chandka Medical College larkana from January 2013 to December 2014.

Material and methods: Twenty cases were enrolled from outpatient of Eye and Skin Department, Chandka Medical College Hospital Larkana. Patients were subjected to detail history, complete eye and skin examination and relevant investigations.

Results: 20 patients, 12(60%) males and 8 (40%) females with xeroderma pigmentosum were recruited in a gender ratio of 1.5 : 1. All patients had skin freckles and increased sensitivity to sun light. Photophobia and conjunctivitis was seen in 18(90%) cases, keratitis and dry eyes was seen in 12 (60%) cases and 4 (20%) cases had malignancies of lids and conjunctiva proved on biopsy. Our cases belongs to larkana and sukkur division.

Conclusion: The present study highlights the eye involvement in xeroderma pigmentosum and thereby early detection of eye disease prevent the malignancies and blindness

Key Words: Xeroderma pigmentosum,—Ocular manifestations

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INTRODUCTION

Xeroderma pigmentosum is a group of rare inherited disorders with autosomal recessive inheritance and is manifested by increased skin sensitivity to ultraviolet (UV) light, abnormal skin pigmentation, photophobia, conjunctivitis and increase tendency to develop skin cancer, especially on sun exposed areas of body.[1] Neurological abnormalities may also be found in some patients. Many enzymatic defects are also observed in these patients[2,3]. According to classification there are eight genetic complementation subgroups of this disorder from XP-A to XP-G and a variant group XP-V [4] with each group having a different gene alteration [5]. Its incidence varies from one region to another. In USA the incidence is 1:250 000 in general population [5]; however it is much higher in some other countries like Japan 1 : 40 000.

There is frequent eye involvement in xeroderma pigmentosum and there is tendency of skin and eye lesions to become malignant in adolescence or early adult life. The skin may become atrophic and ectropion may occur. The bulbar conjunctiva may become thin and atrophic, with inflamed patches resembling phlyctenules.. With infilteration of cornea the patients become photophobic and may also start having blepharospasm.[6,7]

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MATERIALS AND METHODS

This prospective study comprised of 20 patients recruited from two departments, Department of Dermatology and Ophthalmology of Chandka Medical College larkana and was completed in two years period from January 2013 to December 2014. All cases were examined by a dermatologist and a ophthalmologist and clinical diagnosis was made. Complete eye examination like lids, adnexa, anterior segment, posterior segment was done in all cases. Lids and conjuctival incisional biopsy was done in cases where lids and conjuctiva was ulcerated.

RESULTS

There were 12 (60%) males and 8 (40%) females in a ratio of 1.5 : 1. Their ages range from 2 to 26 years with a mean age of 14 years. All cases belonged to Larkana and Sukkur division. All patients showed skin freckles and pigmentation of face and exposed parts of body and sensitivity to sunlight. Ocular manifestations were frequently seen in all cases, like photophobia and conjuctivitis in 18 (90%) cases, keratitis and dry eyes in 14 (60%) cases , lid skin squamous cell carcinoma 2 (10%) cases, basal cell carcinoma 1 (5%) case, conjuctival carcinoma in situ 1(5%) case. All patients were advised to avoid sun light and to apply sun block

whenever needed and were treated accordingly. All patients with eye complaints were given topical ciprofloxacin eye drops 2 hourly, methyl cellulose eye gel 2 hourly during the day times and tobramycin eye ointment at night. 1 patient with conjuctival carcinoma in situ, corneal abscess and perforation was treated by enucleation. 3 patients with lid carcinoma were treated by excisional biopsy and lid reconstruction by plastic surgeon.

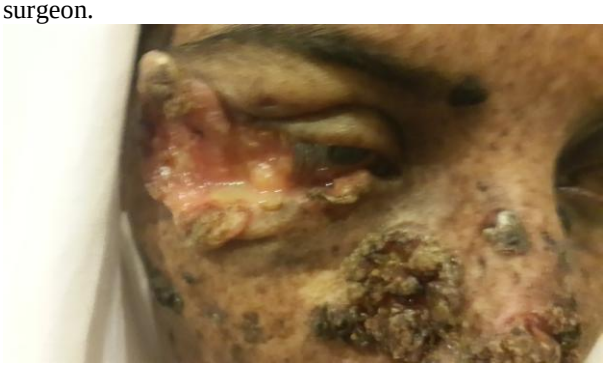

Figure No.1: Basal cell carcinoma of eye lid

Figure No.2: Perforation and endophthalamitis of left eye

DISCUSSION

Defective repair of DNA damaged by ultraviolet radiation play a key role in pathogenesis however there is a great influence of latitude and weather over the symptoms of Xeroderma pigmentosum .[8,9] Pakistan is a tropical country and its climate of varies from one region to another. Eyes are commonly involved in xeroderma pigmentosum. We found photophobia and conjunctivitis in 90% of our patients which is markedly higher than reported by Kraemer *et al* and Brian P *et al* , 21% and 51% of their cases repectively.[10,11] Conjunctival changes such as conjunctivitis, xerosis and congestion was seen in 90% cases. Corneal involvement in xeroderma pigmentosum may lead to dryness, exposure keratitis, opacification, angiogenesis, ulceration and even perforation. In our study corneal involvement was seen in 60% cases which is comparable with the study done by Goyal *et al* [12] who noticed in 40% cases. In our cases 2 (10%) patients showed squamous cell carcinoma and 1 (5%) patient showed basal cell carcinoma of eye lid (Figure 1). One patient showed extensive globe conjunctival carcinoma in situ with secondary corneal abscess, perforation and endophthalamitis of left eye (Figure 2), which is enucleated. In the same patient also there was right eye conjunctival carcinoma in situ, and growth was excised completely. We observed ocular malignancies in 20% cases while another study done by Kraemer et al found in 11% of cases. [13]

CONCLUSION

In our study we conclude that early detection, continuous surveillance and and timely intervention may prevent many complications especially ocular malignancies and blindness in patients with xeroderma pigmentosum.

Conflict of Interest: The study has no conflict of interest to declare by any author.

REFERENCES

1. Lehmann AR, McGibbon D, Sttefainin M. Xeroderma pigmentosum. Orphanet J Rare Dis 2011;6:70.
2. Reed WB, Landing B, Sogarman G, et al. Xeroderma pigmentosum. Cliniacl and laboratory investigations of its basic defect. JAMA 1969; 207:2073-9.
3. Thompson LH. Nucleotide excision repair: in relation to human disease in Dna repair in higher Eukrycts. Vol 2, Torowo NJ. Human press 1998; 335-93
4. Sovoboda DI, Brltey LP, Vosj HM. Defective bypass replication of a leading stand cyctobutane thymine dimer in xeroderma pigmentosum variant cell extract caver Rs 1998;58:1445-8.
5. Robbins JH, Kraemer KH, Luzner MA et al. Xeroderma pigmentosum: an inherited disease with sun sensivity, multiple cutaneous neoplasm and abnormal DNA repair. Ann Intern Med 1974; 80:221-48.
6. Neil JV, Kodai M, Brever R, et al. The incidence of consanguineous matings in japan: with remarks on estimation of comparative gene frequencies and expected rate of appearance of induced recessive mutations. Am J Hum Genet 1949;1:156-78.
7. Theilmann HW, Edler L, Popanda O, et al. Xeroderma pigmentosum patients from the federal repuablic of Germany: decrease in post UV colony-forming ability in 30 Xeroderma pigmentosum fibroblast strands is quantitatively correleted with a

decrease in DNA incising capacity. J Cancer Res Clin Oncol 1985;109:227-40.
8. Clever JE, Carter DM. Xeroderma pigmentosum variants: influence of temperature on DNA repair. J lnvest Dermatol 1973;78:295-7.
9. Takebe H, Nishigori C, satoh Y. Genetics and skin cancer of xeroderma pigmentosum in Japan. Jpn J Cancer Res (Gann) 1987;78:1135-43.
10. Kraemer KH, Slor H. Xeroderma pigmen-tosum. Clin Dermatol 1985;3:33-69.
11. Brian P, Brooks Amy H, Rachel J, Pishop et al, Opthomol 120(7).
12. Goyal JL, Roa VA, Srinvasan R et al. Oculo-cutaneous manisfestation in xeroderma pigmentosum. Br J Opthamol 1994;78:295.
13. Kraemer KH, Lee NN, Scotto J. xeroderma pigmentosum. Cutsneous, Ocular and neurologic abnormalties in 830 published cases. Arch Dermatol 1987; 123:241-50.