Scholars Journal of Applied Medical Sciences (SJAMS)
Abbreviated Key Title: Sch. J. App. Med. Sci.
ISSN 2320-6691 (Online)
ISSN 2347-954X (Print)

A Unit of Scholars Academic and Scientific Society, India
www.saspublishers.com

Ophthalmology

Xeroderma Pigmentosum: Case Series

Dr. Bhavesh Makkar[1*], Dr. Deepak Mishra[2], Dr. Prashant Bhushan[3]
[1]Senior Resident, IMS, BHU, Varanasi, Uttar Pradesh, India
[2]Assistant Professor, IMS, BHU, Varanasi, Uttar Pradesh, India
[3]Associate Professor, IMS, BHU, Varanasi, Uttar Pradesh, India

Case Report

***Corresponding author**
Dr. Bhavesh Makkar

Article History
Received: 11.06.2018
Accepted: 21.06.2018
Published: 30.06.2018

DOI:
10.36347/sjams.2018.v06i06.037

Abstract: Xeroderma Pigmentosum (XP) is a rare autosomal recessive disease. It is caused due to defect in deoxyribonucleic acid (DNA) repair. Eye involvement is seen in 80% of cases. XP patients have intolerance for sunlight which damages the skin as well as the eyes of patients. In extreme cases all exposure to bright sunlight must be avoided. XP patients are also called as Children of the Night. XP patients have freckle like skin pigmentation, multiple cutaneous and ocular malignancies. In late stages neurodegeneration may also occur. XP patients may present with eyelid atrophy or malignancies, corneal dryness, exposure keratopathy and conjunctival tumours. Among the ophthalmic manifestations lid freckles is the most common. We report a case series with almost all typical features in the skin and eyes.
Keywords: Xeroderma Pigmentosum, Lid freckles, cutaneous and ocular malignancies.

INTRODUCTION

Xeroderma pigmentosum is a rare fatal disesase. It is autosomal recessive in inheritance. We report two cases with almost all typical features in the skin and eyes.

CASE REPORT

Two cases one of 12 years old male child and other 15 years old girl was brought to us with complains of bright light intolerance, watering and blurred vision.

History

Both the children have had full term normal delivery. Both belonged to a Hindu family and parents had non consanguiuous marriage.

After the age of six years children started developing roughness of skin surface along with hyperpigmented spots on the exposed parts of the body. The skin lesion gradually started increasing in number and pigmentation. With due course of time whole of the face, forearm and legs were involved. The lesion over the nose developed tenderness and was also associated with occasional bleeding. Six months back both of them developed photophobia which was associated with opaque cornea.

This disease has not been noted in the families of both patients.

General examination

The children were moderately built, non-irritable but resented light. They were mentally mature for their age. All over the face, both forearm and legs had flat hyperpigmented irregular spots varying in sizes from 2-4 mm in diameter. The spots were more concentrated on the extensor aspect of the upper and lower extremities. Both of them developed growth over the anterior aspect of the nose. It was associated with occasional bleed. Teeth were chalky white in colour and widely separated (Figure 1).

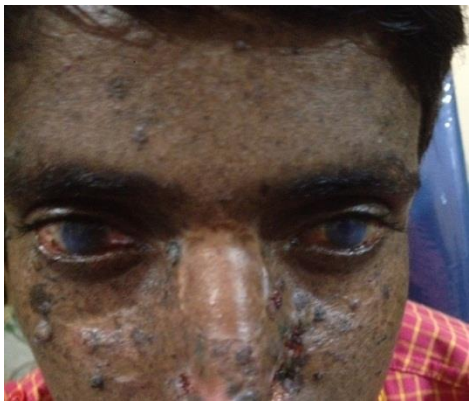

Fig-1

Examination of the central nervous system, respiratory system and cardiovascular system did not

reveal any abnormality. Children were afebrile and had normal appetite.

Ocular examination

First case of 12 year old male child

RIGHT EYE

Both the lids had similar hyperpigmented patches along with lateral lower lid ectropion. Eyelashes were normal in colour and quantity. A horizontal linear tender mass of approx 0.75x0.50 cm was seen at nasal conjunctiva.

A diffuse macular central corneal opacity was seen. Iris and lens were hazily seen and appears normal. Rest details could not be evaluated due to hazy media and photophobia. (Figure 2).

Fig-2

LEFT EYE

Both the lids had similar hyperpigmented patches. Eyelashes were normal. Conjunctiva showed loss of normal vascular pattern. Inferotemporally macular corneal opacity was seen. Iris and lens were hazily seen and appears normal. Rest details could not be evaluated due to hazy media and photophobia. (Figure 3)

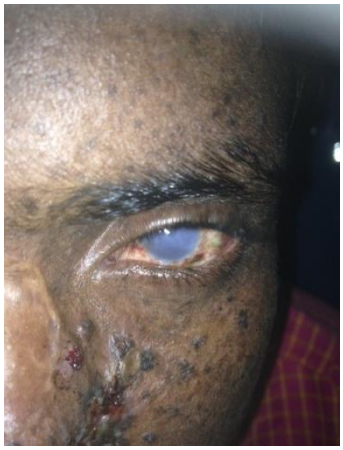

Fig-3

Second case of 15 year old female child

RIGHT EYE

Both the lids had similar hyperpigmented patches. Eyelashes were normal. Conjunctiva showed loss of normal vascular pattern. Macular central corneal opacity was seen. Iris and lens were hazily seen and appears normal. Rest details could not be evaluated due to hazy media and photophobia.

LEFT EYE

The left eye showed the similar appearance as the right eye.

DISCUSSION

XP was first described by a Hungarian dermatologist, Moriz Kaposi [1]. XP is a hereditary disease with autosomal recessive inheritance although sex linked transmission has also been reported. XP has 100% penetrance. Its incidence is not significant in India. It results from mutation in any one of the eight genes responsible for repair of DNA. The first seven genes takes part in nucleotide excision repair (NER) [2].

The genetic defects in XP leads to premature sunlight induced damage including hyperpigmentation, hypopigmentation, lentigos, actinic keratosis and atrophy. Neurological abnormalities are seen in 25% of patients. Neurological abnormalities include neuronal degeneration leading to sensorineuronal hearing loss, ataxia, areflexia, and microcephaly. In our case no neurological deficiency was noted. The neuronal degeneration in XP is due to improper repair of DNA damaged by endogenous metabolites [3].

The most common ocular features noted are blepharospasm and photophobia. The changes that may be seen in the eyelid are erythema, pigmentation, atrophy and malignant changes. Cicatricial ectropion and symblepharon may also occur [4]. Corneal manifestations are exposure keratitis, edema, corneal ulceration and perforation. The manifestations in the conjunctiva are conjunctivitis, pinguecula, symblepharon, melanosis and tumours [5]. Squamous cell carcinoma, malignant melanomas and limbal stem cell deficiency have also been reported. The manifestations in the iris are iritis, stromal atrophy, pigment abnormalities and rarely melanoma. The posterior segment abnormalities are rare as it is protected by cornea and lens. Very rarely choroidal melanoma may develop [6].

Rigorous implementation of protection from UV rays may prolong the life of XP patients. Normal life span is possible in XP patients if they are protected from UV light and do not have any neurological problems [7].

The personal protective measures such as long opaque clothing, sun hats, UV protective sun glasses with side shields and long hair style are useful. All tumors should be adequately excised at the earliest opportunity.

Routine audiometry, measurement of head circumference, assessment of gait and deep tendon reflex testing can usually serve as a screen for the presence of XP-associated neurologic abnormalities. Unfortunately the neurological problems cannot be halted. Management includes hearing aids, speech therapy, occupational therapy and physical therapy.

Surveillance is very important in XP. Vitamin D supplementation is required. Current management of eyelid tumors is complete resection using Mohs' micrographic surgery, with or without reconstruction, or other tissue sparing techniques. Conjunctival tumors should be excised and treated with adjuvant cryotherapy/irradiation/topical chemotherapy [8]. Malignant limbal tumors can be removed by iridocyclectomy or enucleation. Keratoplasty and topical chemotherapy is useful for corneal tumours. Iris tumors may be managed with local excision, plaque radiotherapy, or enucleation [9]. Choroidal melanomas are treated with plaque radiotherapy. For orbital tumours, surgical excision with adjunctive radiation can be therapeutic. The new approach is the introduction of DNA repair enzymes into the skin by means of specially engineered liposomes.

Prenatal diagnosis done by amniocentesis and in vitro cell culture is the only method to prevent this disease. Genetic counselling emphasizing the importance of consanguiuous marriage in the causation of disease is also important.

There are very few cases reported in India. The reporting of every case might help us in knowing the exact incidence and prevalence of XP in India.

REFERENCES

1. Hebra F, Kaposi M. On diseases of the skin including exanthemata. Volume III. New Sydenham Soc. 1874; 61:252–8.
2. Stefanini M, Kraemer KHK. In: Neurocutaneous Diseases. Ruggieri M, Pascual-Castroviejo I, Di Rocco C, editor. Chapter 51. 2008. Xeroderma pigmentosum; pp. 771–792.
3. Robbins JH, Kraemer KH, Merchant SN, Brumback RA. Adult-onset xeroderma pigmentosum neurological disease--observations in an autopsy case. Clinical neuropathology. 2002;21(1):18-23.
4. Reese AB, Wilber IE. The eye manifestations of xeroderma pigmentosum. American Journal of Ophthalmology. 1943 Sep 1;26(9):901-11.
5. Crisp WH. The journal of the Egyptian Medical Association, historical number, volume 11: December, 1928. Cairo. American Journal of Ophthalmology. 1929 May 1;12(5):421-2.
6. Kitagawa KO, Inoue M. Choroidal malignant melanoma occurring in a patient with xeroderma pigmentosum. Folia Ophthalmol Jpn. 1981;32:657-63.
7. Shetty R, Girish BS, Ballal R, Permi HS, Makannavar P, Alva V. Xeroderma pigmentosa with multiple cutaneous malignancies: A rare case report and review of literature. NUJHS. 2013;3:76-8.
8. Kearsley JH, Fitchew RS, Taylor RG. Adjunctive radiotherapy with strontium-90 in the treatment of conjunctival squamous cell carcinoma. International Journal of Radiation Oncology• Biology• Physics. 1988 Mar 1;14(3):435-43.
9. Shields J. Melanocytic tumors of the iris stroma: Intraocular tumors: Atlas and textbook. Chapter 2. 2008.