Case Report

Xeroderma Pigmentosum in a Child: An Early Ocular Manifestation

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Abstract

A 17-month-old boy of first-degree consanguineous parents came with a history of poor vision and photophobia. He also had multiple hyper-pigmented skin lesions all over the body since 8 months of age. The skin lesions were generalized, hyper-pigmented, non-tender, not itchy but photosensitive. Initially, the skin lesion was treated by local physicians but when the lesion was spreading, and subsequently when he had developed visual problems, then visited a pediatrician. His family had no history of a similar type of skin disorder. His developmental milestone was age-appropriate. His eyes were sensitive to sunlight and exposure to light causes redness along with poor vision. There was large corneal diameter, conjunctival xerosis, intense photophobia and poor vision in both eyes. Skin survey revealed hyper-pigmented macules. Hair and nail apparatus were normal. There was no mucosal or dental involvement, no ulceration or growth elsewhere. Hearing was normal. By a multidisciplinary team this case was diagnosed as Xeroderma Pigmentosum clinically. This case was managed by proper counseling advised to protection from sunlight and follow up. We like to share our experience of diagnosing and treating this rare case in a child with early ocular manifestation.

Key words: *Xeroderma Pigmentosum, Early Manifestation*

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Introduction

Xeroderma pigmentosum (XP) is an autosomal recessive, rare pigmentary atrophic childhood disease that can progress to the early development of senile changes in skin of sun-exposed areas. XP was first described by Hebra and Kaposi in 1874. In 1882, Kaposi coined the term xeroderma pigmentosum for the condition, referring to its characteristic dry and pigmented skin. Xeroderma pigmentosum is also known as DeSanctis- Cacchione syndrome. The basic defect underlying the clinical manifestations is a NER (Nucleotide excision repair) defect leading to the defective repair of DNA damaged by UV ray. NER involves the removal and replacement of damaged DNA with new DNA. Two types of NER exist: global GG-NER (Genome NER) and TC-NER (Transcription-coupled NER). In addition, the immunosuppressive effects of UV ray-B radiation may also be involved in the pathogenesis of XP. The estimated frequency of XP in United States is 1:25000 and more in Japan. Its incidence is not significant in the Indian context.[1] XP is a genetic disease characterized by defective repair of damaged DNA. Authors are reported the ophthalmologic manifestations of XP, the ophthalmologic manifestations were present in 62% of cases; where the age of patients was 7 to 22 year. Photophobia were seen in all patient. Patients with XP can develop squamous cell carcinoma at an early age.[2] XP is a hereditary, rare and fatal disease of the skin. Ocular involvement is found about in 80% of cases. A case with typical cutaneous and ocular manifestations is reported.[3] The common abnormalities were found conjunctivitis (51%), corneal neovascularization (44%), dry eye (38%), corneal scarring (26%), ectropion (25%), blepharitis (23%), conjunctival melanosis (20%), and cataracts (14%). Thirteen percent of cases had some degree of visual axis impingement, and 5% of patients had no light perception in one or both eyes. History of ocular surface cancer or ocular surface cancer was present in 10% of cases.[4] The prevalent findings include eyelid changes, observed in 80.9% cases, and ocular surface changes as punctate

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keratopathy (76.2%), corneal neovascularization, and corneal opacities. About 29% presented corneo-conjunctival tumor. More than half of patients had history of treatment of ocular neoplasia. Ocular burning was mostly reported.[5] This paper presents a case study of XP in children. Affected individuals may have a variety of clinical symptoms, which may include problems of the skin and oral mucosa, ocular manifestations, and neurologic impairment. A number of precautions must be taken when treating these cases, which include proper shielding from damaging light.[6] XP with early ocular manifestation are discussed in this report.

Case Presentation

17 month old male of a first degree consanguineous parents came to pediatrics outpatient department of Khwaja Yunus Ali Medical Collge and Hospital, Enayetpur, Sirajgonj, Bangladesh, with the history of poor vision and photophobia especially when sun exposed. The Child often kept bumping with different objects during movement or walking due to poor vision especially in sun light. Mother also complaints of multiple hyper pigmented skin lesions all over the body, more on the sun exposed parts mostly face, scalp and upper trunk sparing the palm and sole; beginning from 8 months of his age, Initially the rash was erythematous macular rash. His mother described the skin lesion were generalized, hyper pigmented area, non-tender, not itchy but photosensitive. There was no history of similar type of skin disorder in his family. His birth history was uneventful with having proper feeding practices. He was immunized as per EPI schedule. His developmental millstone is age appropriate. Initially the skin lesion was treated by local physicians but when the lesion was spreading, and poor vision, then visit to pediatrician. His eyes were sensitive to sunlight and exposure to light causes redness along with poor vision. After taking ophthalmologist consultation, there is large corneal diameter (13mm), conjunctival xerosis, intense photophobia and poor vision in both eyes (Fig 2). By a multidisciplinary team involving a Pediatrician, Dermatologist and Ophthalmologist we diagnosed him as a case of Xeroderma Pigmentosum clinically. He was playful. Vital signs were normal. OFC was 49 cm which lies on 50th centile. Anthropometry revealed that his weight was 8.5 kg, length 78 cm, weight for length on 25th centile and length for age on 25tn centile weight for age -3SD. Skin survey revealed hyper pigmented macules distributed over face, scalp, upper trunk and extremities (Fig 1) sparing the palm and sole. Hair and nail apparatus were normal. There was no mucosal or dental involvement, no ulceration or growth elsewhere. Hearing was normal. Others revealed no abnormality. His Hb 11.5gm%, TC 8000/cmm, Neutrophil 46%, Lymphocytes 53%, Platelets 2,40,000/cmm; serum IgE- 70 IU/ml. Now, he is being under regular follow up by a multidisciplinary team involving a Pediatrician, Dermatologist and Ophthalmologist. Advised to avoid sun exposer, use glass and observe of any changes in lesions. Till writing this case report our case did not develop any further complications.

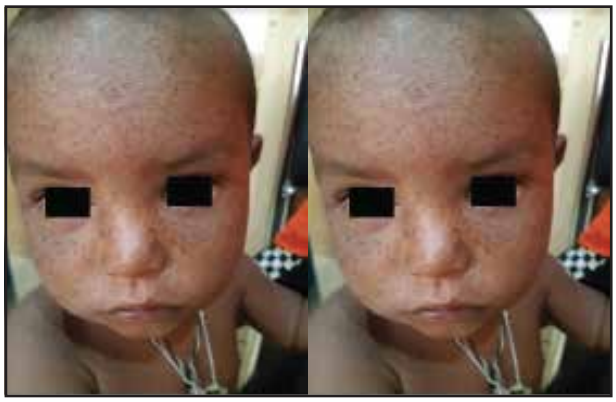

Figure 1: Pigmentary changes in Xeroderma Pigmentosum.

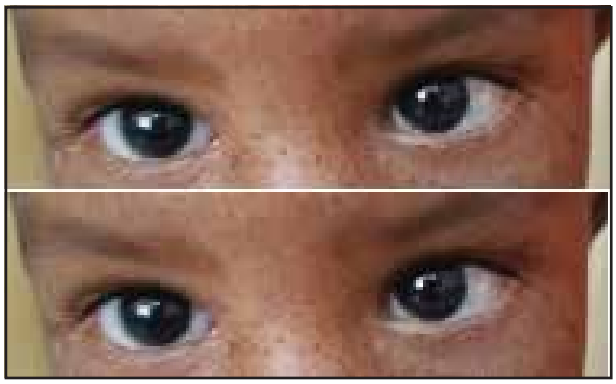

Figure 2: Eye changes in Xeroderma Pigmentosum

Discussion

Both sexes are almost equally affected. History of consanguinity found in 30% of cases of affected children.1 In our case it was a consanguineous marriage and the child was males. Another reported, a case of XP in three consecutive siblings of a Nigerian family where both male and female were affected.[7] Authors reported that the diagnosis can be made in the first year of life; [8] where our case at 17 month of age. Our patients started developing skin rash around 6 months of age. Significant corneal scarring and multiple cutaneous skin lesions observed in sun-exposed areas in a case of an 8 year-old girl with neuro-developmental delay had been present since 3 months of age, and whole clinical picture was consistent with XP.[9]

Report of a 6-year-old boy came with skin lesions all around the body since birth. No parental consanguity. No similar illness in the family. Normal growth. The chief complaints were photophobia and defective vision.[10] that's complaints were similar to our case presentation.

In this case report, patients had intense photophobia, conjunctival dryness with poor vision. Ophthalmologic abnormalities are usually limited to the anterior, UV- exposed portion of the eyes: conjunctiva, cornea and lids found in another case report.[11] In a case series of seven patients with XP found dry eye in 100 % and conjuctival melanosis in 50% cases in study abroad.[12] There is no cure for XP. Avoidance of sun exposure is obligatory, including the use of sunscreens, long-sleeved protective

clothing, wide-brimmed hats and UV-absorbing eye glasses when outdoors. All sources of UV radiation in the home, school or work environment should be identified and eliminated, if possible. Individuals should be taught to recognize new lesions and monitor for any changes, including size and color, in pre-existing skin lesions for early detection of cancer. Genetic counseling should be offered for families at risk. Genetic testing are not available in our country as well as at abroad everywhere.

Conclusion

Xeroderma Pigmentosum is rarely diagnosed at the remote area in Bangladesh. This case is being reported to create awareness about XP among the root level doctors of rural Bangladesh. More important is the psychological and social support to the patient and family. Because of the awful appearance people are scared of these patients. People should be made aware that this is not an infectious or contagious disease and genetic counseling should be provide to avoid consanguineous marriages.

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Multidisiciplinary approach always ameliorate the diagnosis and treatment of a rare case. Special thanks to Dermatologist and Opthalmologist of Khwaja Yunus Ali Medical College and Hospital.

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