CASE REPORT

TWO SIBLINGS WITH XERODERMA PIGMENTOSUM, AFAR, ETHIOPIA

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ABSTRACT

Xeroderma Pigmentosum is one of the rare autosomal-recessive skin lesions with a broad variety of clinical manifestations that lead to cell hypersensitivity to ultraviolet radiation. In addition to extensive skin and mucosal damage with malignant transformation, it is characterized with ocular involvement and progressive neurologic deterioration. We mention the clinical characteristics of siblings xeroderma pigmentosum come to our pediatric unit, Dubti General Hospital, Afar, in two adolescent siblings from Asab, Eretria.

Key words: *Xeroderma Pigmentosum, Siblings, Afar*

INTRODUCTION

Xeroderma pigmentosum (XP) is a rare autosomal recessive DNA repair condition, a wide range of clinical presentations leading to ultraviolet radiation cell hypersensitivity, actinic skin damage. It is also associated with cancer of the eyes and mouth of the UV-exposed areas of the skin and mucous membranes, and progressive neurological degeneration (1,2). Ocular involvement is reported in 40-50 percent of cases, and neurological complications arise in about 30 percent of cases and can be serious (1-3). We mention the clinical characteristics of siblings in this paper, in which we found them classic Xeroderma pigmentosum cases.

Case 1

A 12-year-old male patient has had scaly hyperpigmented skin lesions since birth. He also has vision problems in sunny areas and hearing difficulties. His caregivers complained that he had much less school operation and growth than his peers. In addition, they complained there is decreased hearing ability. Otherwise no history of drug use, injury, yellowish discoloration and itching. No parental consanguinity was reported. No similar illness in the family except in his younger sister. His birth history was normal; delivered with spontaneous vaginal delivery and no circumstance.

During physical examination, he had scaly hypopigmented skin lesions across his face, trunk, and extremities. Oral lesions with encircling the mouth were also present. There were also eye injuries including heavy tears and corneal cloudiness. In addition, on neurologic assessment motor, sensory and reflexes were normal yet memory and arithmetic capacity were deranged.

On investigation his complete blood count, PITC is unremarkable. Other investigations of organ function test, ESR, abdominal and chest ultrasound were normal. Histologic examination was not possible due to lack of availability of the service.

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Fig 1. Frontal picture of the face showing hyperpigmented skin lesions with thickening and ulceration. Injected conjunctiva and bilateral corneal cloudiness. Oral ulceration is also depicted.

Case 2

A 9-year-old female sister of him presented with scaly skin lesions with hyperpigmentation and freckling. She also had also a wound which has stayed for more than 3 years over her scalp. She was incapable of learning due to the skin lesion which hurts her from attending school and family complains has hearing difficulty and mental retardation. Otherwise no history of abnormal body movement and loss of consciousness any time. No other family history of similar illness.

On physical examination have scaly skin lesions over the chicks, fungating mass over the scalp with pus and putrid odor. She had difficulty of hearing and vision.

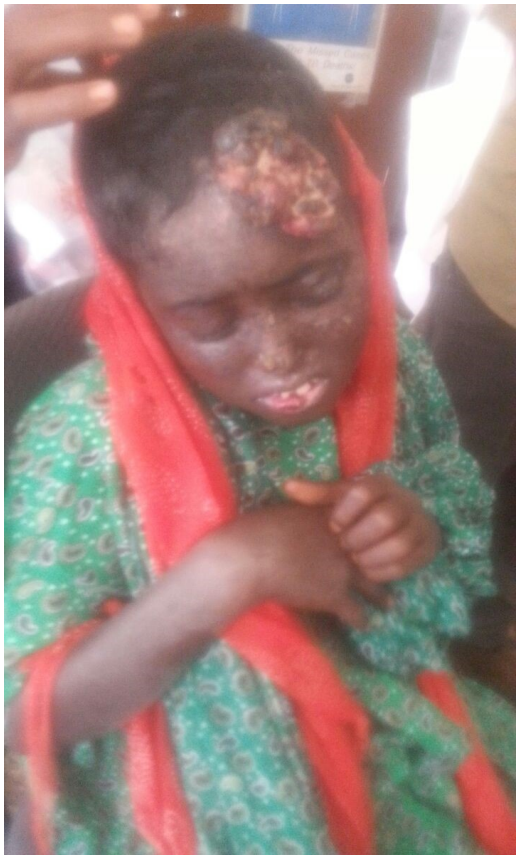

Fig 2. Hyper pigmented skin lesions over the face. Fungating mass with ulceration over the frontal area of left forehead.

On investigation she has unremarkable CBC results and RBS. Screening for HIV and echocardiograph was done both non revealing results. Other investigations of organ function test, ESR, abdominal and chest ultrasound were normal. Histologic setup was not available the diagnosis of xeroderma pigmentosum was made by clinical constellation of sign symptoms. They were managed with conservative management to avoid sun exposure, wound care and referred for better management.

DISCUSSION

Xeroderma pigmentosum affects both genders equally presenting with wide spectrum of disease manifestation. Most of the sign and symptoms occur after few minutes of sun exposure with hypersensitive response to the skin. extreme sunburn after a few minutes of sun exposure with freckling, dryness and pigmentation due to degenerative and proliferative cutaneous changes, including hyperpigmentation, skin atrophy, poikiloderma, actinic keratosis during childhood (2,4,5). Skin changes might transform to basal cell carcinoma (BCC), squamous cell carcinoma (SCC) and melanoma.

Both of our patients displayed photophobia a nd corneal opacification bilaterally with impa ired vision for which locomotive assistant is r equired. In Xeroderma Pigmentosum patients have acute / chronic blepharitis with blepharospasm and photophobia are common symptoms. Eyelid alterations with exposure to keratitis, corneal opacification, and pterygia are common (2,3,4).

One of the most common causes of pain and discomfort in feeding, breathing is actinic cheiltis. The oral manifestations are usually related to the presence of malignant tumors in the lips, tongue and mucosa of the mouth. We have seen, similarly, that our patients have trouble feeding. In addition, there can also be neurological problems, including hearing loss, impaired balance, cognitive capacity loss, and epilepsy (2-6).

Patients have higher risk of skin cancer like squamous cell carcinoma, basal cell carcinoma and malignant melanoma (7,8,9). Diagnosis is with clinical presentation and genetic testing to detect mutations of nine genes responsible with additional biopsy, ophthalmic evaluation and oncologic evaluation might be needed (1,2,4,6,7)

Treatment of xeroderma pigmentosum is avoidance of the UV light. Protective clothing, sunglasses and sunscreen are the mainstay of treatment despite no cure. Vitamin D supplementation and retinoic acid treatment are important means of management in halting the disease progress. Cutaneous neoplasia is generally treated with curettage or by excision (8-10).

CONCLUSION

Early detection of the disease improves the quality of life of xeroderma pigmentosum patients. As much as possible sun exposure must be avoided in such patients with an eye to the emergence of skin and brain cancers. Rehabilitative therapy also plays an important role as patients do have intellectual impairment and ophthalmic evaluation is recommended. Genetic screening is also required with regard to our cases.

Consent for publication

The authors have agreed to publish at your journal site with agreement of all.

Availability of data and material

The original card of the patients can be retrieved form the card store of the hospital in any time.

Conflicts of interest

The authors declare no conflict of interest.

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REFERENCES

1. DiGiovanna, J. J. and K. H. Kraemer (2012). "Shining a light on xeroderma pigmentosum." Journal of investigative dermatology 132(3): 785-96.
2. Pandey, A., K. Kuldeep, et al. (2013). "Xeroderma pigmentosa with ocular association: case report." Case Rep Clin Med 2: 466-69.
3. Halpern, J., B. Hopping, et al. (2008). "Photosensitivity, corneal scarring and developmental delay: Xeroderma Pigmentosum in a tropical country." Cases journal 1(1): 254.
4. Pathy, S., K. Naik, et al. (2005). "Squamous cell carcinoma of face with xeroderma pigmentosa: a case report." Indian Journal of Medical and Paediatric Oncology 26(1): 47.
5. Feller, L., N. Wood, et al. (2010). "Xeroderma pigmentosum: a case report and review of the literature." J prev med hyg 51(2): 87-91.
6. Jasuja, S., A. Mathur, et al. (2003). "Xeroderma pigmentosum with squamous cell carcinoma of face: A rare case report of two siblings." Clin Infect Dis 37: 820-27.
7. Butt, F. M., J. R. Moshi, et al. (2010). "Xeroderma pigmentosum: a review and case series." Journal of Cranio-Maxillofacial Surgery 38(7): 534-37.
8. Kondoh, M., M. Ueda, et al. (1994). "Siblings with xeroderma pigmentosum complementation group A with different skin cancer development: importance of sun protection at an early age." Journal of the American Academy of Dermatology 31(6): 993-996.
9. Chaudhary, M., S. Jajoo, et al. (2012). "Xeroderma Pigmentosum: a case report of two siblings." J Immunodefic Disor 1: 2.
10. Naik, S. M., A. M. Shenoy, et al. (2013). "Cutaneous malignancies in xeroderma pigmentosum: earlier management improves survival." Indian Journal of Otolaryngology and Head & Neck Surgery 65(2): 162-67.