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Photo Essay *Cornea & Refractive*

Corneal manifestations of xeroderma pigmentosa in a heavily pigmented patient

Baltaj Sandhur[1], Charles Zhang[2], John H. Lillvis[2]

[1]Department of Ophthalmology, Jacobs School of Medicine and Biomedical Sciences, [2]Department of Ophthalmology, Ross Eye Institute, Buffalo, United States.

***Corresponding author:**
Baltaj Sandhur,
Department of Ophthalmology, Jacobs School of Medicine and Biomedical Sciences, Buffalo, United States.

baltajsa@buffalo.edu

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A 15-year-old Somali immigrant female presented to the clinic with diffuse hyperpigmented macules [Figure 1a]. Slit-lamp examination demonstrated scattered pigmentation through the conjunctiva and cornea with a dense inferior stromal scar with a large feeder vessel extending nasally [Figure 1b]. The contralateral eye demonstrated similar but less prominent findings [Figure 1c]. Anterior segment optical coherence tomography demonstrated normal epithelium and hyperreflectivity of the anterior stroma consistent with scarring [Figure 1d]. The patient has a strong family history of xeroderma pigmentosa, which includes her three siblings and was subsequently diagnosed with the same disease by dermatology.

Xeroderma pigmentosa is a rare autosomal recessive disease with a prevalence of 1:1,000,000 in the United States with other populations such as Japan, North Africa, and Pakistan having a higher prevalence.[1-3] This disease is caused by a genetic mutation in a variety of genes (*XPA*, *XPB*, *XPC*, *XPD*, *XPE*, *XPF*, *XPG, ERCC1, CSA, CSB, GTF2H5,* and *TTDN1*). These altered genes result in a defective nucleotide excision repair, which is responsible for repairing the DNA following damage from ultraviolet (UV) radiation.[4]

In these patients, mutations in DNA repair caused by UV radiation result in inflammation and neoplasia in many sun-exposed areas, such as the skin and eye.[5] These patients present normally at birth, and depending on the type of mutation, they can present in various ways, from having an exaggerated response to UV radiation causing burning and blistering with minimal exposure versus a normal acute response to UV exposure. Despite these differences, all of them develop pigmented changes in sun-exposed areas, which can present as hypopigmentation, hyperpigmentation, atrophy, and telangiectasias, typically before the age of 2.[4,6]

Ocular disease is evident in almost 40% of patients and affects both males and females equally.[7] Xeroderma pigmentosa most commonly affects the anterior surface of the eye with many experiencing photosensitivity. One study found that the most common abnormalities included conjunctivitis (51%), corneal neovascularization (44%), dry eyes (38%), corneal scarring (26%), and blepharitis (23%). At present, there is no definitive treatment for xeroderma pigmentosa. The patients are advised to avoid sun exposure. The main cause of death in these patients is neurologic degeneration or cancer.[8] Increased awareness regarding the disease, as well as frequent checkups, can improve life expectancy for these patients.[3] This patient retains 20/40 vision and is monitored on a close schedule every three months. The patient was advised to avoid sun exposure to reduce further complications.

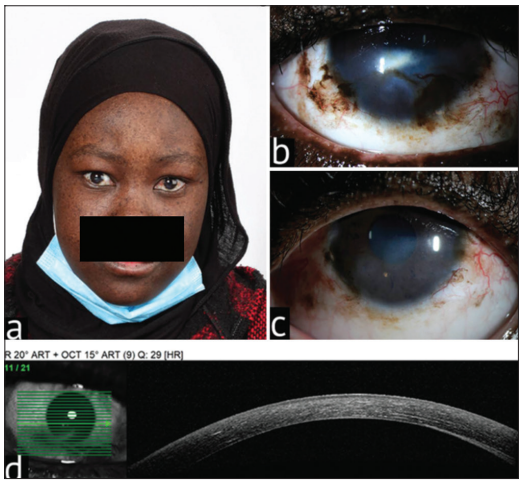

Figure 1: (a) A 15-year-old xeroderma pigmentosa patient with hyperpigmented macules. (b) External slit lamp photograph with inferior stromal scarring along with a feeder vessel in the right eye. (c) External slit lamp photograph with similar findings in the left eye. (d) Anterior segment optical coherence tomography photograph of the right eye with normal epithelium and hyperreflectivity of the anterior stroma.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Fassihi H, Sethi M, Fawcett H, Wing J, Chandler N, Mohammed S, *et al.* Deep phenotyping of 89 xeroderma pigmentosum patients reveals unexpected heterogeneity dependent on the precise molecular defect. Proc Natl Acad Sci U S A 2016;113:E1236-45.
2. Lehmann AR, McGibbon D, Stefanini M. Xeroderma pigmentosum. Orphanet J Rare Dis 2011;6:70.
3. Leung AK, Barankin B, Lam JM, Leong KF, Hon KL. Xeroderma pigmentosum: An updated review. Drugs Context 2022;11:2022-2-5.
4. Black JO. Xeroderma pigmentosum. Head Neck Pathol 2016;10:139-44.
5. Brooks BP, Thompson AH, Bishop RJ, Clayton JA, Chan CC, Tsilou ET, *et al.* Ocular manifestations of xeroderma pigmentosum: Long-term follow-up highlights the role of DNA repair in protection from sun damage. Ophthalmology 2013;120:1324-36.
6. DiGiovanna JJ, Kraemer KH. Shining a light on xeroderma pigmentosum. J Invest Dermatol 2012;132:785-96.
7. Ramkumar HL, Brooks BP, Cao X, Tamura D, DiGiovanna JJ, Kraemer KH, *et al.* Ophthalmic manifestations and histopathology of xeroderma pigmentosum: Two clinicopathological cases and a review of the literature. Surv Ophthalmol 2011;56:348-61.
8. Kraemer KH, DiGiovanna JJ, Tamura D. Xeroderma pigmentosum. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJ, *et al.*, eds. GeneReviews(®). Seattle, WA: University of Washington; 1993-2024.

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