Photosensitive Malar Rash in an Infant

Apoorva Sharma, Keshavamurthy Vinay

Department of Dermatology, Venereology and Leprology, Postgraduate Institute of Medical Education and Research, Chandigarh, India

Xeroderma pigmentosum (XP) type A in an infant

A 1-year-old girl presented for evaluation of a photosensitive rash, which was present since early infancy. The parents complained of sunburn of the face and inconsolable cry on exposure to sunlight. There was also a tendency of the child to keep her eyes closed during sunlight exposure. There was no other relevant family or developmental history. Examination revealed well to ill-defined erythematous plaque covering the malar area and bridge of the nose, extending to the temples with brownish scaling and light to dark-brown hyperpigmentation [Figure 1]. Whole exome sequencing was performed, which revealed a homozygous 2 base pair deletion in exon 5 of the xeroderma pigmentosum type A (XPA) gene resulting in a frameshift mutation and premature truncation of the protein 3 amino acids downstream to codon 217 (p.Lys217 GlufsTer3). Based on history, clinical examination, and genetic testing, diagnosis of xeroderma pigmentosum type A was made. Parents were counseled about strict sun protection and possible complications of the disease in the form of skin cancers and neurological involvement. One month after strict photoprotection and proper usage of sunscreen, the rash resolved with freckles and post inflammatory hyperpigmentation.

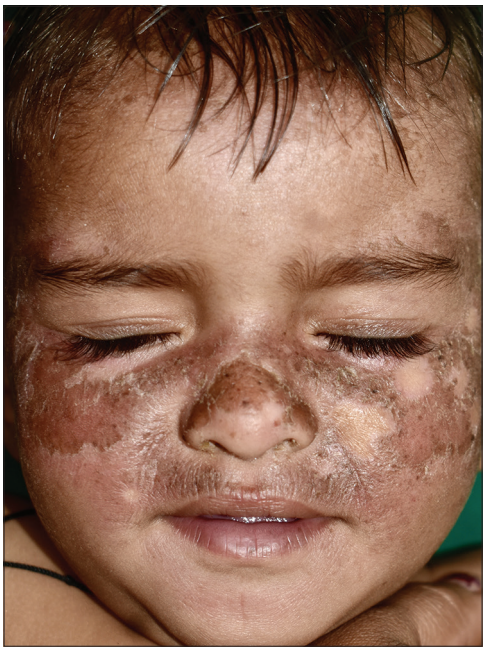

Figure 1: Well to ill-defined erythematous plaques with brownish scaling and light to dark-brown hyperpigmented macules involving the malar area and dorsum of the nose

Xeroderma pigmentosum is a rare autosomal recessive condition marked by heightened sensitivity to sunlight, pigmentation that resembles freckles, and a significantly higher incidence of skin cancer.[1] The prognosis differs by the type of XP, and XPA patients have maximum risk of developing neurological complications. Disorders with photosensitivity in an infant include XP, Rothmund–Thomson syndrome (RTS), Bloom syndrome, Cockayne syndrome (CS), lupus erythematosus, and Hartnup's disease. Bloom syndrome is characterized by dwarfism with photosensitive telangiectatic erythema in the malar area of the face, and can be quite similar to RTS and XP, in the first 2 years of life.[2] It is possible to distinguish Bloom from RTS due to the earlier onset and the presence of hypoplastic/absent thumbs and a variety of other skeletal abnormalities including increased risk of having osteosarcoma in the latter.[3] Another variant of XP is the XP-CS overlap. Although traditionally CS is known, not to have any predisposition to malignancies. In a study by Natale *et al*., it was found that all 40 patients showed lab-confirmed photosensitivity, and the majority of them burned readily in the sun; skin cancer before age 10 was seen (skin cancer is less common in XP-CS patients) but may occur very early in life.[4] The constellation of clinical findings of a

How to cite this article: Sharma A, Vinay K. Photosensitive malar rash in an infant. Indian Dermatol Online J 2024;15:562-3.

Received: 20-Jul-2023. **Revised:** 19-Aug-2023. **Accepted:** 15-Sep-2023. **Published:** 03-Apr-2024.

Address for correspondence:
Dr. Keshavamurthy Vinay, Department of Dermatology, Venereology and Leprology, Postgraduate Institute of Medical Education and Research, Sector 12, Chandigarh - 160 012, India. E-mail: vinay.keshavmurthy@gmail.com

Access this article online

Website: https://journals.lww.com/idoj

DOI: 10.4103/idoj.idoj_556_23

Quick Response Code:

particular syndrome might not be completely seen at the time of first clinical presentation. It is hence essential to perform genetic sequencing for counseling and surveillance of complications.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Acknowledgement

We would like to acknowledge Dr. Akash P Mustari for his assistance in clinical photography of the patient.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Sun Z, Zhang J, Guo Y, Ni C, Liang J, Cheng R, *et al*. Genotype-phenotype correlation of xeroderma pigmentosum in a Chinese Han population. Br J Dermatol 2015;172:1096-102.
2. Arora H, Chacon AH, Choudhary S, McLeod MP, Meshkov L, Nouri K, *et al*. Bloom syndrome. Int J Dermatol 2014;53:798-802.
3. Sharma R, Lewis S, Wlodarski MW. DNA repair syndromes and cancer: Insights into genetics and phenotype patterns. Front Pediatr 2020;8:570084.
4. Natale V, Raquer H. Xeroderma pigmentosum-cockayne syndrome complex. Orphanet J Rare Dis 2017;12:65.