Bilateral ocular neoplasia in a young boy with mild facial freckles

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DESCRIPTION

An 11-year-old boy presented to the outpatient department of a tertiary eye hospital with progressively growing raised pigmented lesions temporally in both eyes (BE) for 2 years. He consulted elsewhere 6 months prior where excision of Left Eye (LE) lesion was performed suspecting pterygium. Histopathological examination was not done there. The lesion recurred soon after at the same location as reported by the patient. He gave history of mild photosensitivity around malar area. He was Indian by ethnicity and was born out of a non-consanguineous marriage with two unaffected siblings. Blood profile showed no evidence of immune compromise. Mild freckling only in periocular and malar area was noted (figure 1). Irregularly raised mass with pigmentation and keratin along with feeder vessels were noted in the temporal bulbar conjunctiva in BE (figure 2A,B). Uncorrected distance visual acuity in BE was 20/20. With clinical picture suggestive of ocular surface squamous neoplasia (OSSN), conjunctival tumour excision with alcohol kerato-epitheliectomy, cryotherapy and placement of Dulbecco's Modified Eagle's medium preserved human amniotic membrane graft (AMG) was performed for BE. Taking 4-mm margin around the tumour resulted in a conjunctival defect of 11×7 mm in BE, which necessitated AMG for conjunctival closure. Histopathological examination from the right eye (RE) revealed moderate dysplasia with dysplastic cells in lower two-thirds of epithelium (figure 2C). Superior, inferior and temporal margins along with the base was noted to be free of dysplasia and atypia. The histopathological examination of LE showed hyperplastic squamous epithelium with atypical cells with epithelial downgrowth and breach in the basement membrane continuity along with focal invasion into the stroma, thus confirming the diagnosis of invasive OSSN (figure 2D). Base, superior, inferior and temporal margins of the LE were also free of tumour invasion. Postoperatively, the AMG was in situ with no recurrence or residual lesion in BE at 1-month follow-up (figures 1 and 2E,F). The nature of illness was explained to the patient and parents, while keeping him on close follow-up to rule out recurrence. Dermatology and paediatric consultations were sought for systemic evaluation and ruling out other skin and oral malignancies.

BE OSSN is extremely rare in immunocompetent children.[1] It has been seen in association with xeroderma pigmentosum (XP) in this group of individuals.[2] Classical XP is an autosomal recessive disorder which presents as early as first few weeks of life with extreme photosensitivity and lentiginosis or freckles in sun-exposed areas.[3] By adolescence, the disease is classically advanced with multiple skin, ocular and oral malignancies, and dry atrophic pigmented skin with extreme photosensitivity.[3] It is mainly diagnosed clinically and confirmed by genetic analysis.[3]

In the absence of the typical features of XP, occurrence of BE OSSN in a young boy, as in our case, with minimal facial freckles and mild photosensitivity is suggestive of a rare milder form of XP.

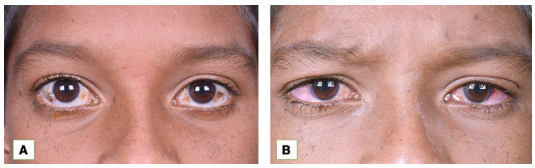

Figure 1 (A) External photograph of the face showing mild freckles in the periocular region and malar area with pigmented raised lesions in the temporal bulbar conjunctiva in both eyes (BE). (B) Postoperative external photograph at 1 month showing amniotic membrane graft in place in BE with no recurrence or residual lesion.

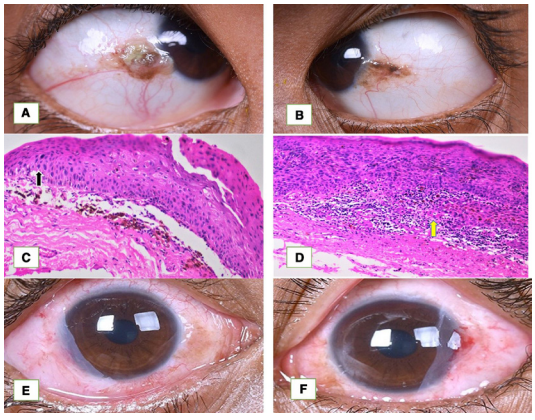

Figure 2 (A) Right eye (RE) showing gelatinous pigmented lesion in the temporal bulbar conjunctiva with extensive keratin and (B) left eye (LE) showing gelatinous pigmented lesion in the temporal bulbar conjunctiva with a speck of keratin. Histopathological examination of the excised lesion showing (C) dysplastic cells in lower two-thirds of epithelium (black arrow) suggestive of moderate dysplasia in RE (H&E, ×100) and (D) invasive squamous cell carcinoma with epithelial downgrowth and invasion into the stroma (yellow arrow) in LE (H&E, ×100). Postoperative clinical picture of the (E) RE and (F) LE at 1 month showing a well-placed amniotic membrane graft with no recurrence or residual lesion in BE.

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This explains the repeated misdiagnosis of a potentially life-threatening condition before the patient presented to us. This case demonstrates the importance of diagnosing this serious ocular and systemic condition especially in the absence of typical dermatological features.

Learning points

- Bilateral ocular surface squamous neoplasia in an immunocompetent child without typical dermatological features may be a rare presentation of atypical form of xeroderma pigmentosum.
- Management includes surgical conjunctival tumour excision with alcohol kerato-epitheliectomy, cryotherapy and amniotic membrane graft with close follow-up to rule out any recurrence.

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