Diagnosis and management of SUFU mutation associated Gorlin syndrome in a kidney transplant recipient

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Learning Objectives

- Gorlin syndrome (GS) or basal cell nevus syndrome is characterized by calcification of the falx cerebri, multiple basal cell carcinoma (BCC), craniofacial features, palmoplantar pits, and various fibromas[1-2].
- GS with rare SUFU mutations (≤5%) can be challenging to diagnose due to subtle clinical presentations and the absence of PTCH mutations.[2]
- Broadened genetic testing can also be a useful tool to diagnose GS in those with negative PTCH mutations.
- Early diagnosis of GS in transplant patients is crucial, as it can impact immunosuppressive strategies.

Case Presentation

A 30-year-old female with a history of kidney transplant due to reflux nephropathy, medulloblastoma, meningioblastoma, and ovarian fibroma, presented to the dermatology clinic for a transplant skin check.

- Skin examination per **Figure 1 & 2**.
- Relevant physical exam findings: subtle pits on palms, and widely spaced teeth
- No personal history of skin cancer and uncertain family cancer history
- Shave biopsies from an eyelid lesion (Fig 1), the scalp (Fig 2), and the upper back (Fig 3) revealed basal cell carcinoma (BCC).
- Patient was prescribed imiquimod cream for eyelid lesions without improvement.
- Genetic testing showed a mutation in tumor suppressor SUFU gene. (Negative PTCH1, PTCH2, PTEN)

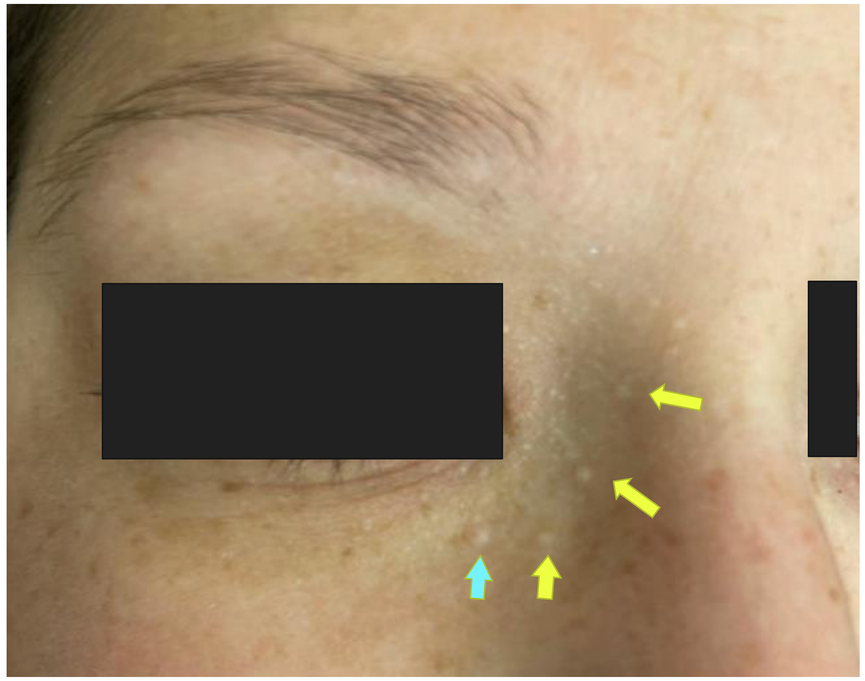

Figure 1: Numerous pearly umbilicated papules on the eyelids and nasal ala. BCC indicated by the light blue arrow.

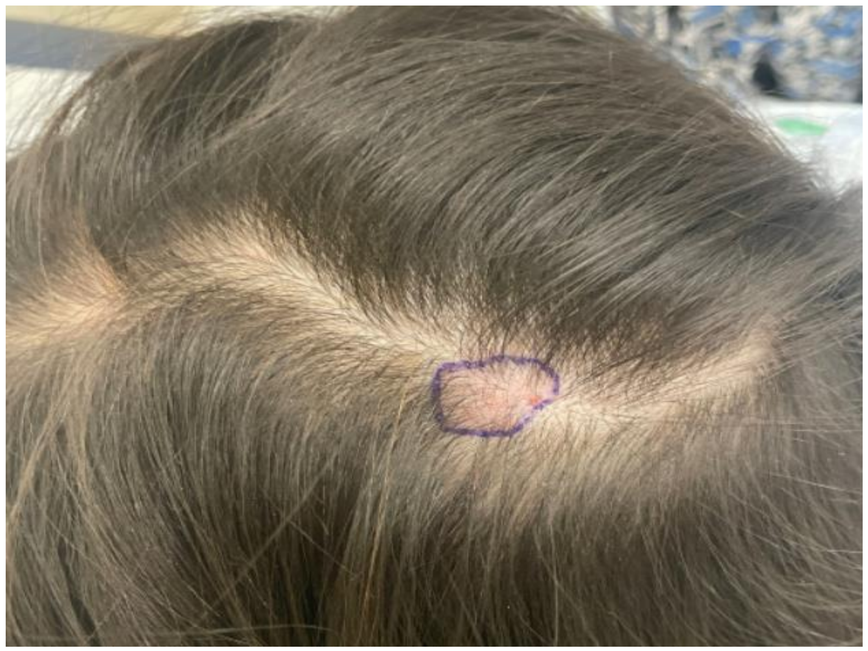

Figure 2. Pink plaque with a few areas of pigment on the left vertex of the scalp.

Outcomes and discussion

- BCCs of the face and upper back removed via shave biopsies and scalp BCC was removed by Moh's surgery.
- Patient referred to photodynamic therapy for periorbital basal cell carcinomas.
- Immunosuppression dosage was reduced and oral niacinamide was added to the regimen.
- The majority of GS cases are associated with PTCH1 mutations, but approximately 5% of cases are due to SUFU mutations.[3]
- SUFU variants have higher risk of developing medulloblastoma, meningioma or ovarian fibroma, and less likely to have jaw cysts, compared to PTCH1 variants.[3]
- GS in transplant patients are understudied, only one case reported in 2009.[4]
- Negative PTCH test should not rule out Gorlin syndrome, especially in patients with multiple BCCs, relevant medical histories and physical examinations specific for GS.
- Cancer predisposition syndrome, including GS, has major implications regarding decision for transplant, immunosuppressive regimen, and ongoing malignancy surveillance/management.

References

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Disclosure

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