

# Gorlin Syndrome Alliance Patient Registry Yields Insights on Patients with Multiple Childhood Basal Cell Carcinomas

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**Introduction:** The Gorlin Syndrome Alliance Patient Registry (GSAPR) includes patient-entered detailed clinical information that provides clinicians and researchers with important insights on rare disease.

**Methods:** A retrospective review of clinical features, prior treatments, and patient preferences for management strategies was conducted through GSAPR.

**Results:** To date, there are 18 participants enrolled in the GSAPR who have indicated their first basal cell carcinoma (BCC) manifested before age 18 years. Of these respondents, 15 reported a diagnosis of 6 or more BCCs, with eight patients reporting over 500 BCCs and two patients reporting diagnosis of over 1000 BCCs. Two patients are presented in detail (Figures 1 and 2).

**Conclusion:** The GSAPR provides a mechanism for investigators to request data access to address clinical questions and to advance understanding of rare disease through patient-derived data.



**Figure 1:** A 4-year-old patient with numerous papules present on the lower eyelids concerning for basaloid follicular hamartoma. No significant change with tacrolimus, and some clinical improvement with sirolimus 1% cream.



**Figure 2:** A 16-year-old patient who has tried numerous treatments:

- Pulsed dye laser
- trichloroacetic acid 85%
- Electrodesiccation and Curettage
- 5-fluorouracil cream
- Imiquimod cream
- Sirolimus 3% cream

Sirolimus has clinically stabilized periorbital BCC's, but lesions on trunk are increasing in number and size.

**Contact:** Collaborators interested in Gorlin Syndrome research are welcome to connect with us after meeting.  
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**RESEARCH OPPORTUNITY:**  
Gorlin Syndrome Alliance  
Patient Registry (GSAPR)



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