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## Pediatric basal cell carcinoma burden and management preferences in Gorlin syndrome: A survey study

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**Pediatric basal cell carcinoma burden and management preferences in Gorlin syndrome: A survey study**



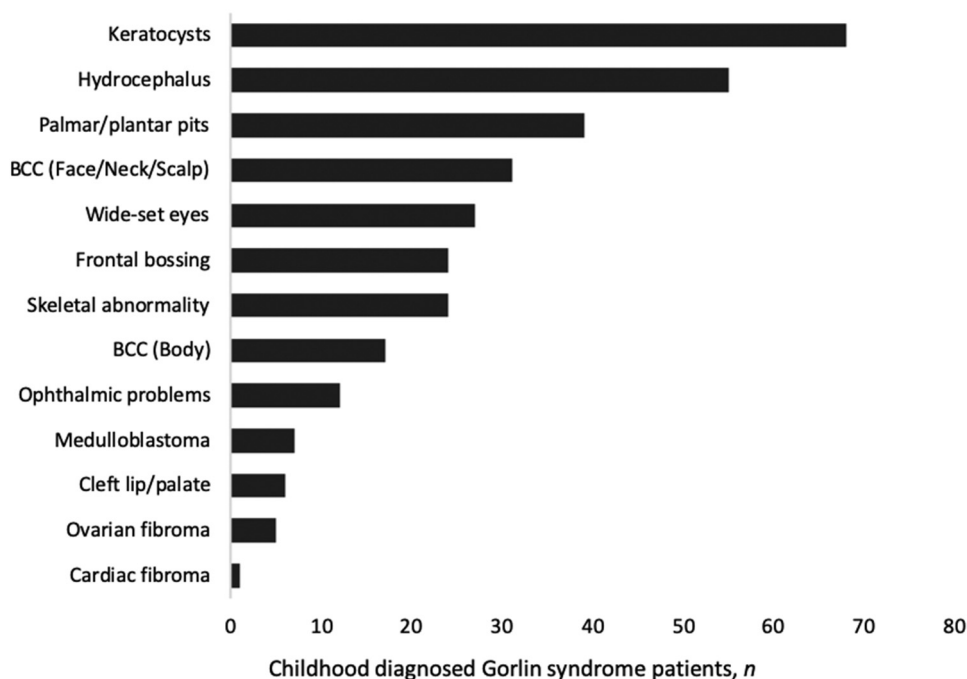
*To the Editor:* Gorlin syndrome (GS) is a risk factor for early basal cell carcinomas (BCCs),<sup>1</sup> although its prevalence of fewer than 1 in 30,000 individuals<sup>2</sup> limits existing literature. There are sparse pediatric GS studies beyond case reports, creating a knowledge gap regarding childhood cutaneous findings and sequelae, including BCC age at onset, quantity, treatments, and impact. Herein, we describe a global survey to illustrate the clinical presentation, childhood perspectives, and BCC management trends for pediatric GS to improve the understanding and inform patient care.

Institutional review board approval was obtained from Massachusetts General Hospital to study survey data collected by the GS Alliance and GS Group.

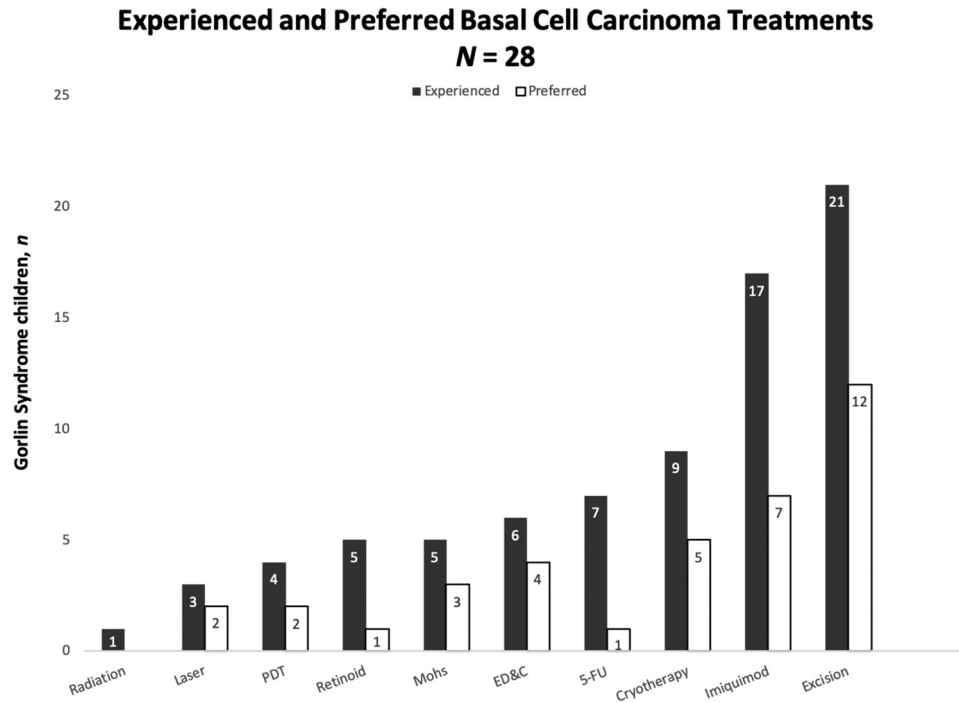
Inclusion criteria for analyses were respondents with GS who were physician-diagnosed in childhood (at ages,  $\leq 18$  years), of whom, 122 qualified. The majority were female ( $n = 93$ , 76.2%), aged 5 to 14 years at diagnosis ( $n = 66$ , 54.1%), White ( $n = 108$ , 88.5%), and born in the United States ( $n = 76$ , 62.3%). Of known mutation status, PTCH1 was reported in 65 of 66 individuals (98.5%) and SUFU was reported in 1 individual (1.5%). The most common presenting characteristics (Fig 1) were keratocysts ( $n = 68$ , 55.7%), large/abnormal skull ( $n = 55$ , 45.1%), palmar/plantar pits ( $n = 39$ , 32.0%), and BCC ( $n = 37$ , 30.3%).

At the time of survey completion, 42 respondents represented children (aged  $\leq 18$  years), of whom 32 (76.2%) had a history of BCC. The 32 children with BCC (65.6% aged  $\leq 13$  years; range, 6-18 years) had notable BCC burden: their cumulative number of tumors was 1 to 30 in 17 patients (53.1%), 31 to 100 in 5 (15.6%), and  $> 100$  BCCs in 10 children (31.3%).

**Presenting Childhood Signs**  
**N = 122**



**Fig 1.** Presenting signs for patients with Gorlin syndrome who were diagnosed at ages of 18 years or younger. Respondents could select more than 1 presenting sign. BCC, Basal cell carcinoma.



**Fig 2.** Basal cell carcinoma treatment modalities that were experienced by patients with Gorlin syndrome aged 18 years and younger with basal cell carcinoma have been shown (solid bars). Treatment modalities that were preferred by the children with Gorlin syndrome with basal cell carcinoma who had experienced the respective treatment have also been represented (white bars). ED&C, Electrodesiccation and curettage; 5-FU, 5-fluorouracil; PDT, photodynamic therapy.

Excision was the most commonly pursued treatment for pediatric BCC ( $n = 21$ ) and preferred by 12 patients who experienced it (57.1%). However, a higher percentage expressed preference for Mohs micrographic surgery (3 of 5), electrodesiccation and curettage (4 of 6), and laser modalities (2 of 3) (Fig 2). The top 10 burdensome aspects of treatments were as follows: time spent off work/school to recover ( $n = 22$ ), time spent on appointments ( $n = 21$ ), time spent traveling to appointments ( $n = 21$ ), mental well-being ( $n = 21$ ), scarring ( $n = 20$ ), physical appearance ( $n = 20$ ), healing time ( $n = 19$ ), pain ( $n = 18$ ), physical well-being ( $n = 18$ ), and quality of life ( $n = 16$ ).

This childhood GS study identifies meaningful trends, demonstrating how patient voices may inform and improve the management of GS. Although literature estimates the cumulative incidence of BCC by the early 20s in patients with GS to be 12% to 50%,<sup>3,4</sup> our data reflect a considerably higher burden with over three-quarters of pediatric respondents (aged  $\leq 18$  years) reporting a history of BCC. Dermatologists, as well as dentists, oral surgeons, and other clinicians, have an

important duty to recognize presenting features of GS for appropriate care. BCC management in children may create treatment challenges, thus a pediatric-centered approach and consideration of patient and caregiver preferences must be used to provide individualized care.<sup>5</sup>

Further research is required to confirm patient-reported data. Results are limited by the unknown response rate and bias of participants who are engaged in patient advocacy organizations and may exhibit a more severe disease expression.

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**Conflicts of interest**

Author Breneiser is the executive director and a board member for the Gorlin Syndrome Alliance. Dr Hawryluk is a board member for the Gorlin Syndrome Alliance (uncompensated). Author Neale has no conflicts of interest to declare.

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