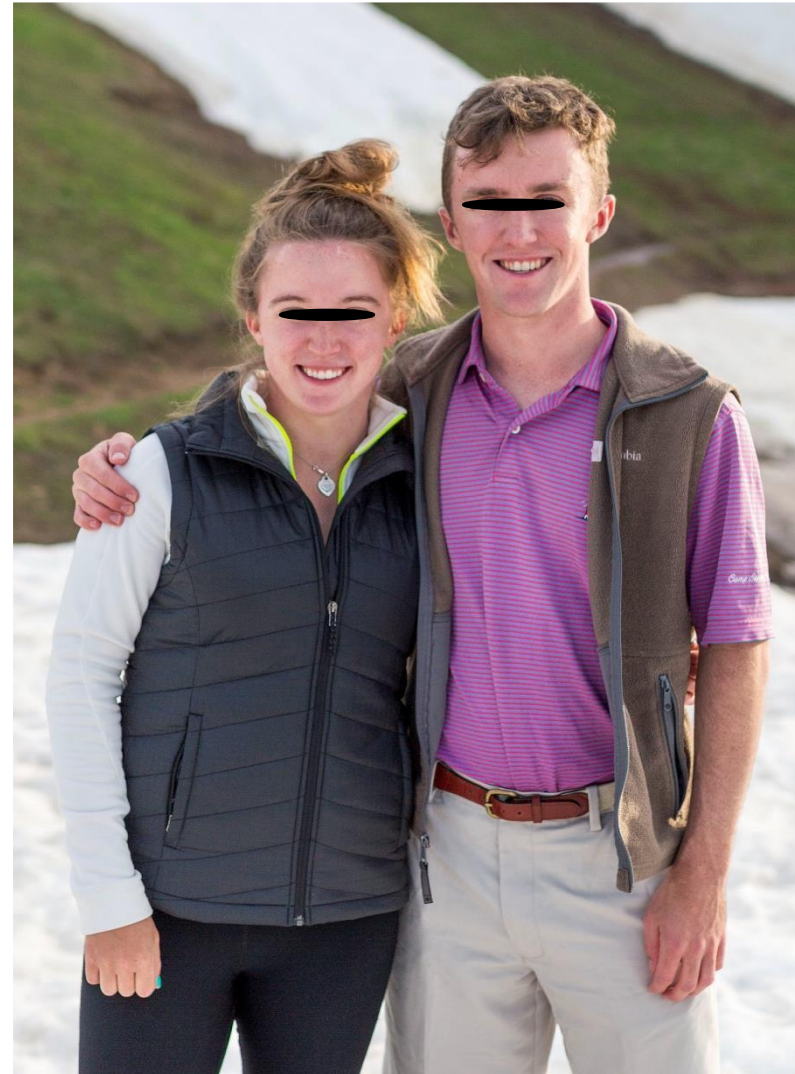


Julie Breneiser, Executive Director

Gorlin Syndrome Alliance

Affected mother of two adults with Gorlin Syndrome

Retired Physician Assistant



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gorlin
SYNDROME ALLIANCE



People with Gorlin Syndrome are Warriors.

We **NEED** you to help fight a multitude of battles!
Care of Gorlin syndrome patients takes a **platoon**.
It requires **lifelong relationships** with *many* providers.

Specialist	Frequency of Visits	Manifestation
Dermatologist	3-6 months <i>throughout life</i>	<i>BCCs</i>
Oral Surgeon	<i>At least every 2 years throughout life</i>	Odontogenic Keratocysts (OKC aka KCOT)
Dentist / Orthodontist	Every 6 months <i>throughout life</i>	Monitoring
Geneticist	Annually for first few years after dx then prn	Early dx & family planning
Mental health provider	As needed. Be mindful of the <i>invisible</i> scars.	Overall disease burden
Others: Neurosurgery, gynecology, cardiology, ENT, ophthalmologist, ENT, orthopedics, PT, OT, ST	As needed depending on manifestations. Gyn visits should start at onset of menses.	Hydrocephalus, macrocephaly, cranial synostosis, calcified ovarian & cardiac fibromas, cleft lip / palate, glaucoma, cataracts, boney anomalies,...
Gorlin Syndrome Alliance (gorlinsyndrome.org)	Essential for individual support and vetted information	Gorlin syndrome

45% of respondents have had
>50 BCCs on their face / neck / scalp.

42% respondents have had
>50 BCCs on other parts of their body

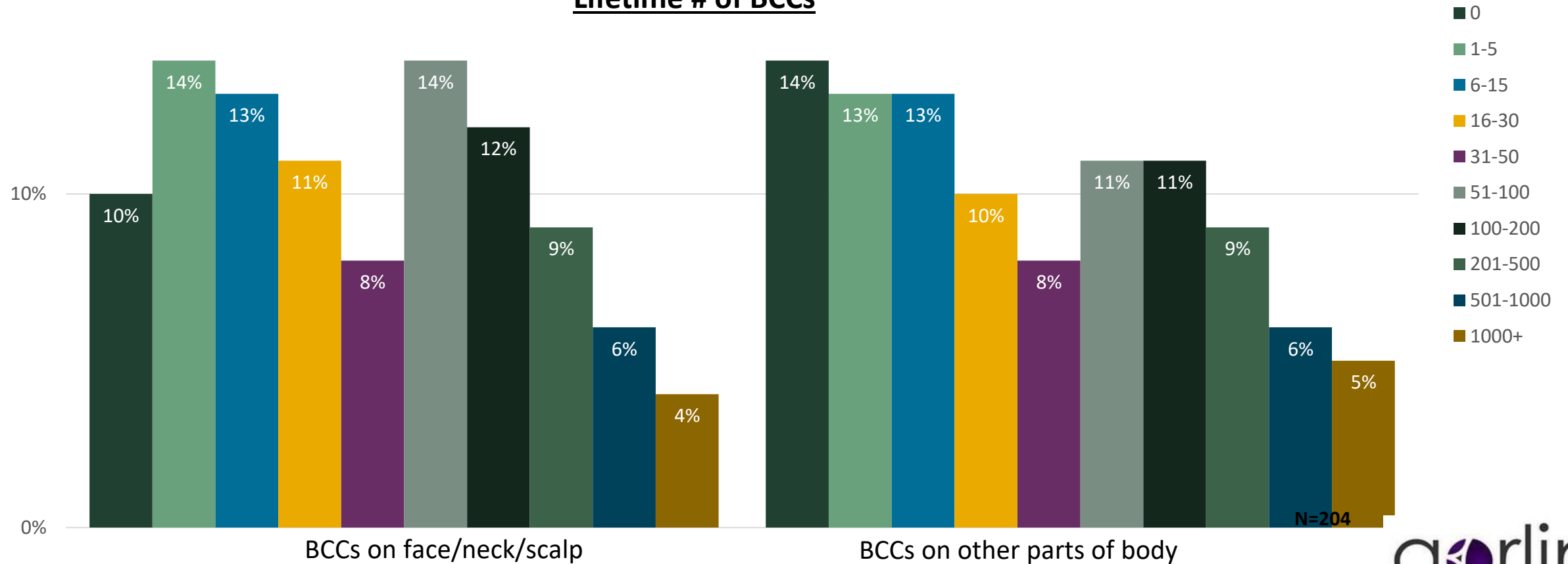
5% had OVER 1,000

20%

10%

0%

Lifetime # of BCCs

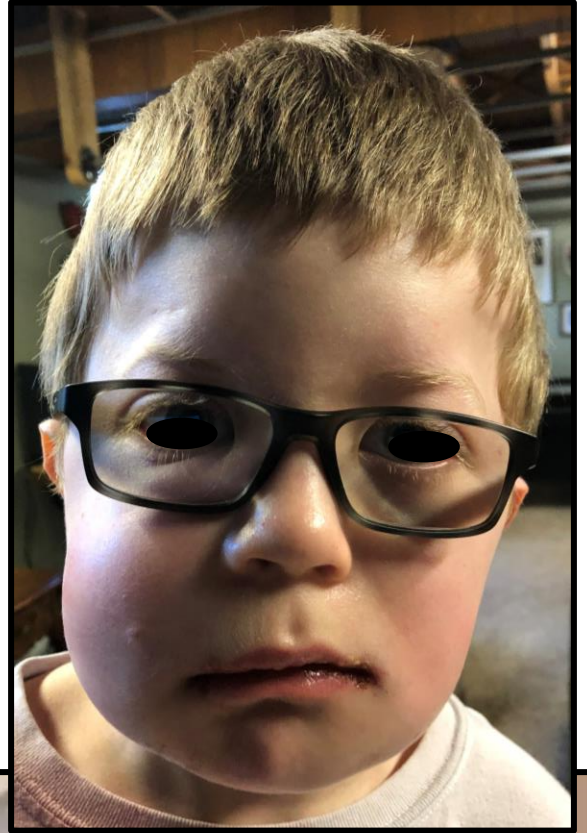




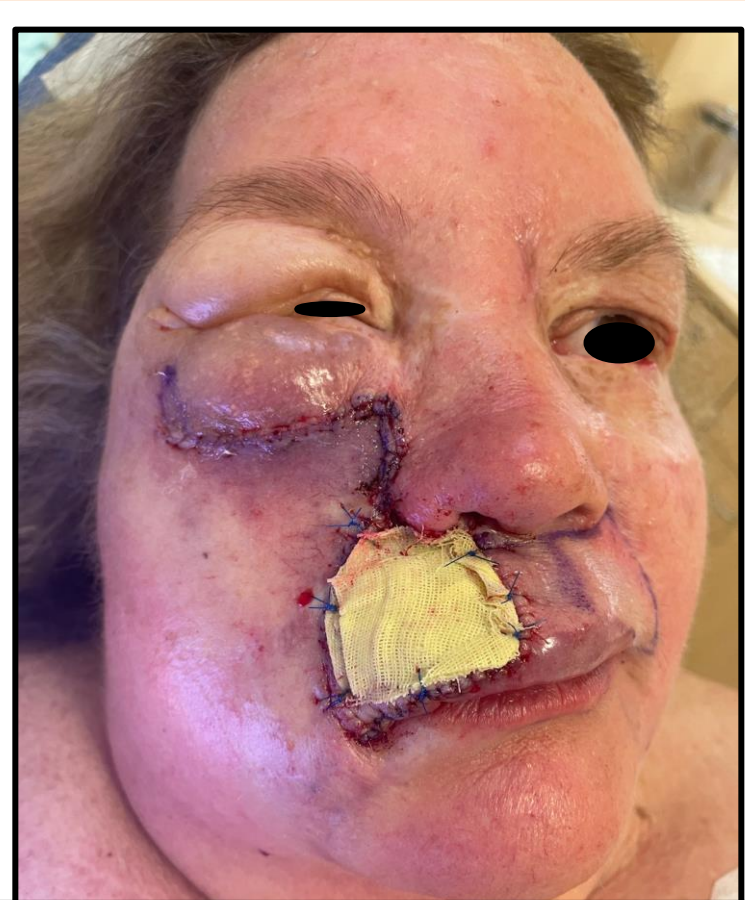
At what age was your
first BCC diagnosed?

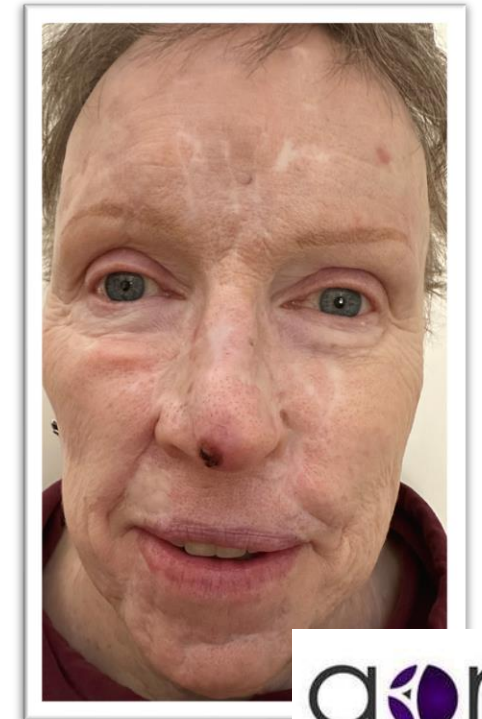
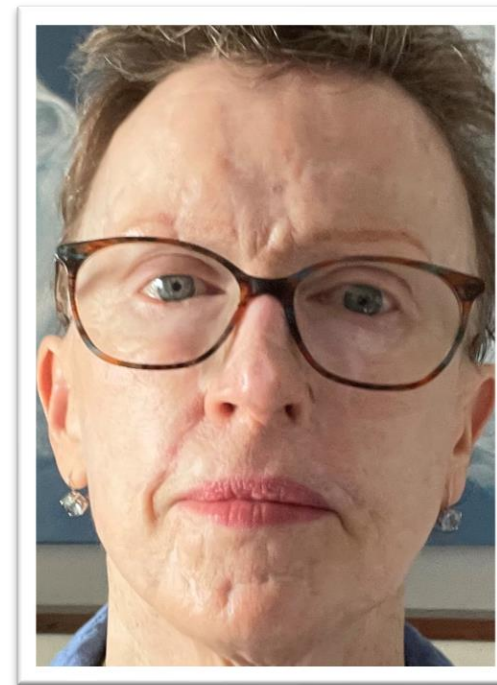
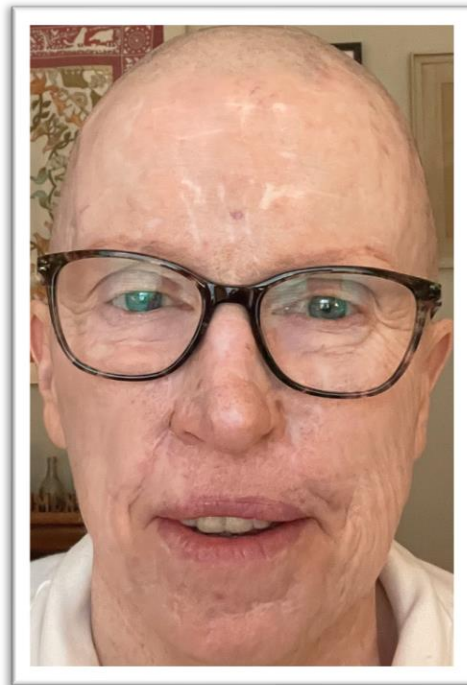
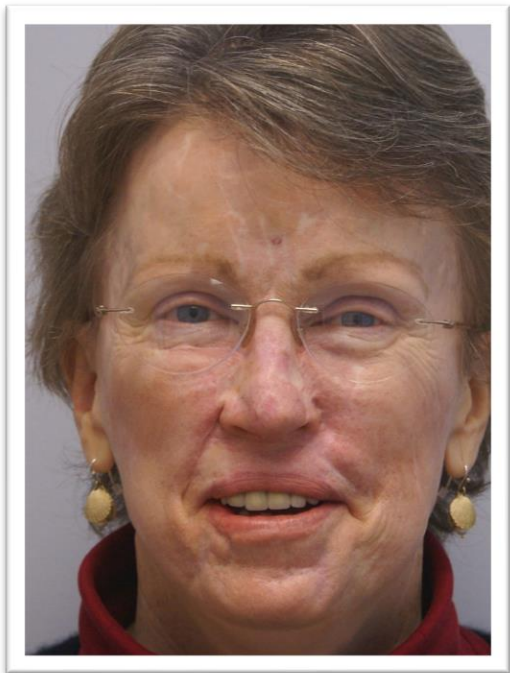
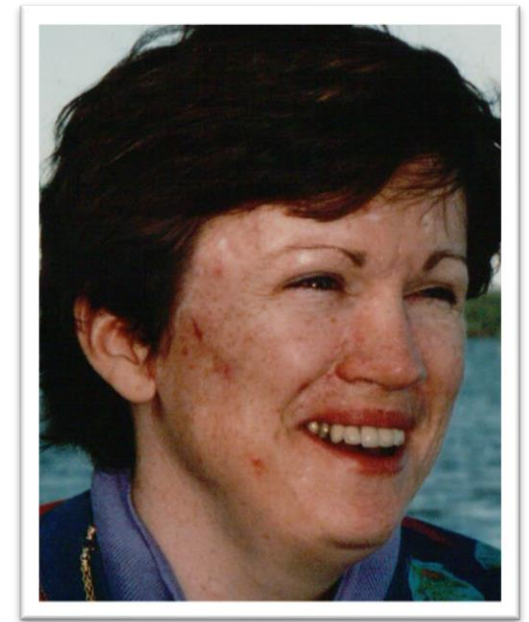
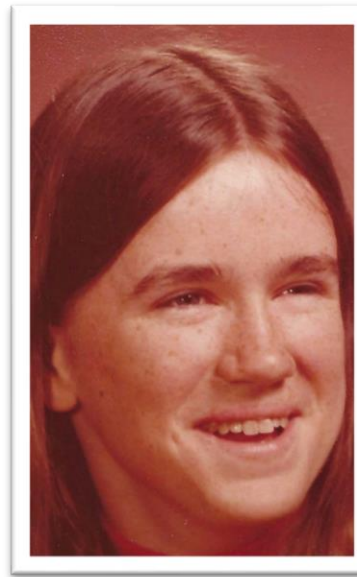
- 0-24 months – 3%
- 2-5 years old – 10%
- 6-10 years old – 23 %
- 11-20 years old – 50%
- 21 and older – 15%











How can *you* help?

- Put yourself in our shoes & be our orchestra conductor.
- Be aware that many deny their disease, avoid and / or burn out on treatment of their BCCs.
- Gently encourage us to take care of ourselves by:
 - a) Following up regularly with appropriate specialists.
 - b) Discussing treatment options, *including clinical trials*.
 - c) Offer a break from treatments but, put an endpoint on this by scheduling next appointment after a certain amount of time off.
 - d) Suggesting daily application of sunscreen. The best kind is the one that is worn every day, clouds or shine. Remind us to wear sun protective clothing & sunglasses.
 - e) Put us on your schedule at the end of the workday or book a double time slot if possible.

How can you help?

- Tell your patients, families, their care partners about the Gorlin Syndrome Alliance: gorlinsyndrome.org

We provide:

- a) Comprehensive website with vetted information. This includes a printable, patient-facing information pamphlet in English & Spanish.
- b) Ongoing patient registry / natural history study. Please *encourage people to enroll* to assist in advancing science leading to better treatments and a cure.
- c) Personal support via individual contacts, virtual community group meetings and town halls, Facebook groups (main page is public, all others are private for affected individuals / families only), Instagram and LinkedIn posts, informative monthly newsletters.
- d) Ask them to join our database. It is FREE, and personal information is not shared without individual consent.
- e) Bi-annual in-person conferences. (June 2025 – Orlando, FL)
- f) Information on clinical trials.

Gorlin Syndrome Alliance Fact Sheet

- Founded in 2000.
- The **mission** of the Gorlin Syndrome Alliance is to thoughtfully support, comprehensively educate, and aggressively seek the best treatments and a cure for those affected by Gorlin syndrome.
- Three pillars to our work: Provide **information**, provide **support**, involvement in **research** by assisting pharma with clinical trials, our own small program that funds research.
- Four staff. Shoe-string budget. “Small and mighty!”
- Eleven-member fully engaged Board of Directors.
- Ten-member Medical & Scientific Advisory Committee with over 290 combined years in practice.
- Conducted a Listening Session and Externally-Led Patient Focused Drug Development meeting for the FDA in 2020 and 2021.
- Executive Director cycled off the FDA’s Patient Engagement Collaborative in the end of 2023.



THANK YOU!



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GSA
Website



GSA
Patient Registry