# *Lifelong Disfigurement and Anxiety Associated with Gorlin Syndrome, a Rare Disease Causing by a Mutation in PTCH1 Leading to Frequent Cancerous Skin Lesions*

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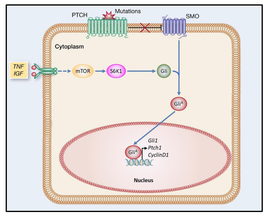
**BACKGROUND:** Gorlin syndrome is most often caused by a genetic mutation of the tumor suppressor "patched" (PTCH1) gene that is typically inherited but may also arise spontaneously.




BCCs in Gorlin syndrome may develop in very early in life (infancy) and occur as frequentlyas 200 times per year.

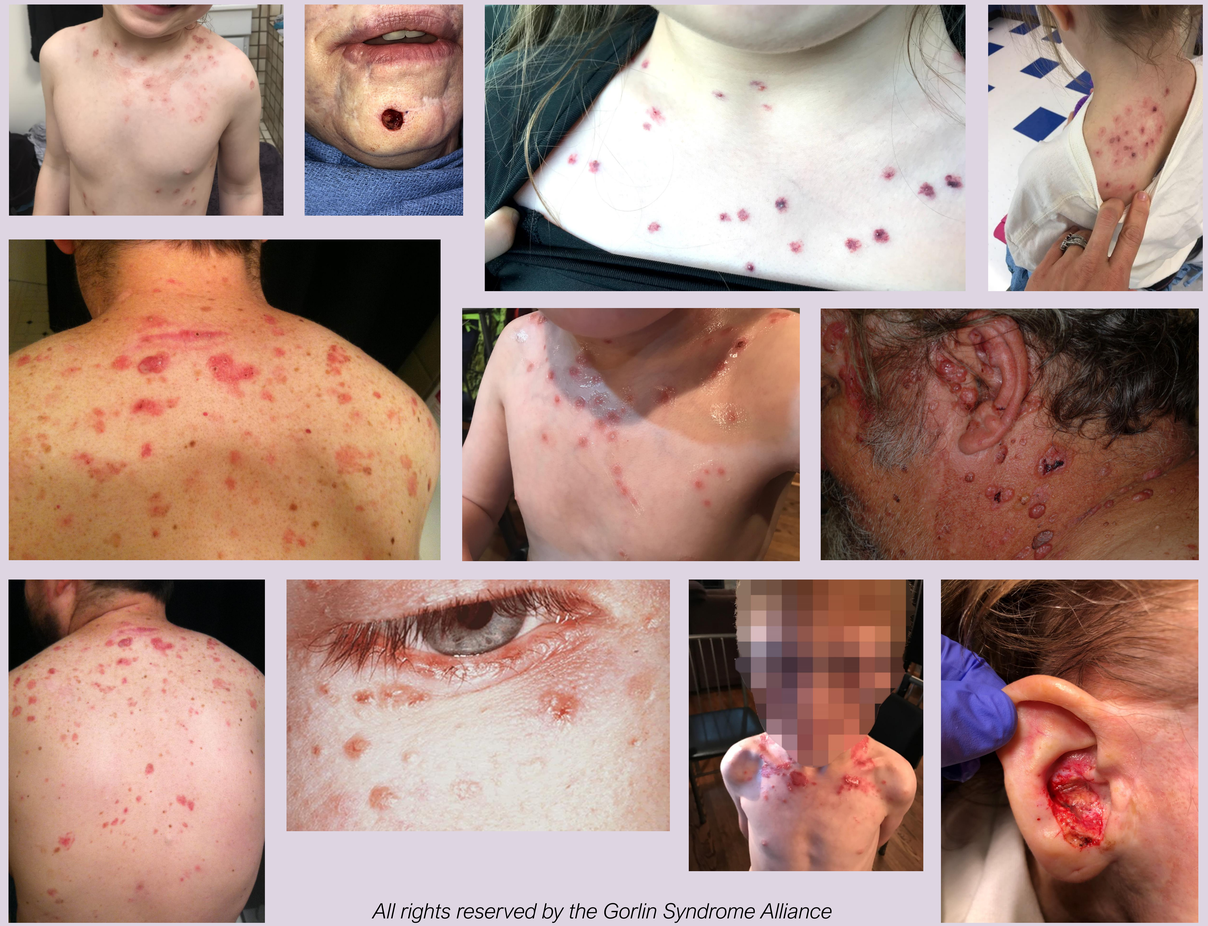
Early diagnosis of Gorlin syndrome is important due to the development of BCCs at an young age and the need to avoid exposure to ionizing and UV radiation, which can exacerbate the condition.

Because Gorlin syndrome can manifest in every organ system, the physical and emotional toll it places on affected individuals and their families is tremendous. Beyond this, the financial burden (medical insurance, lost time form work/school, costs of travel to appointments, etc.) negatively impacts the lives of all involved.

**PLANNED PROGRAM:** The Gorlin Syndrome Alliance and Palvella Therapeutics are collaborating on a planned Phase 2 study to test an experimental targeted treatment, PTX-367 (QTORIN™ rapamycin), in patients with Gorlin syndrome with surgically eligible BCCs. Participants will be identified in part via the Gorlin Syndrome Alliance Patient Registry.

Currently there are no FDA-approved treatments for Gorlin syndrome.

# Individuals of All Ages With Gorlin Syndrome Experience Lifelong Anxiety From Managing Pervasive Cancerous Lesions and Frequent Disfiguring Surgeries



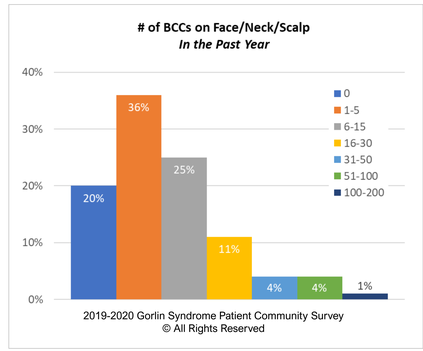


Information on the Gorlin Syndrome Alliance

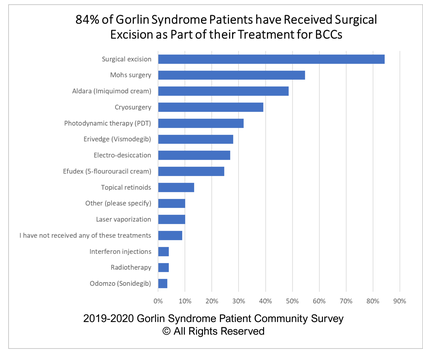
Information on Palvella Therapeutics

## GORLIN CHARACTERISTICS:

- Gorlin syndrome affects ~11,000 people in the United States.




- Radiotherapy is contraindicated in Gorlin patients, leaving surgical excision the standard of care.




- A survey of adults with Gorlin syndrome (*Shah 2011*) shows that 50% have significant depressive symptomatology (using the CES-D scale).