Rich and Full Lives

Skillful, informed management of Gorlin syndrome will give you the freedom to enjoy a rich, full life. The GSA is here to help anyone who has Gorlin syndrome or cares about someone with the condition. For symptoms that may be recurrent and progressive, such as cancerous BCC skin lesions and jaw cysts, we can help you expertly manage your healthcare. The utilization of a knowledgeable medical team along with diligent personal care enables individuals with Gorlin syndrome to thrive.

gorlin
SYNDROME ALLIANCE

The mission of the Gorlin Syndrome Alliance is to thoughtfully support, comprehensively educate, and aggressively seek treatments, with the ultimate goal of finding a cure.

The GSA works to achieve its goals using the three strategic pillars of education, support and research. Our community includes affected individuals, families, friends, medical professionals and researchers.

Learn More About Gorlin syndrome at www.gorlinsyndrome.org

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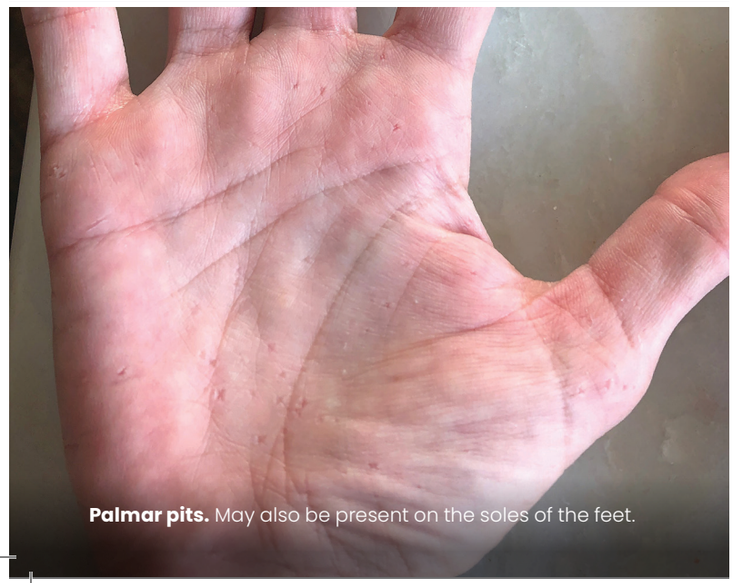
What is Gorlin Syndrome?

Gorlin syndrome (also known as Nevoid Basal Cell Carcinoma syndrome, Gorlin-Goltz syndrome or Basal Cell Carcinoma Nevus syndrome) is a rare genetic disorder. It can affect many parts of the body including the skin, eyes, reproductive system, bones, heart and brain. Individuals with Gorlin syndrome are at increased risk of developing cancers and non-cancerous tumors. It is estimated that 11,000 people in the US live with this syndrome.

Gorlin syndrome is usually caused by a mutation of PTCH, a tumor suppressing gene. (There are less common mutations that may cause the syndrome as well.) The PTCH gene signals to cells when to stop growing and dividing. In Gorlin syndrome, the PTCH gene does not function properly, causing cells to proliferate and form tumors. Gorlin syndrome can be inherited from a parent with the disease or caused by a spontaneous genetic mutation in individuals with no family history. In both of these instances, Gorlin syndrome is present from birth. Individuals with the disease have a 50% chance of passing it to their children.



Palmar pits. May also be present on the soles of the feet.

Symptoms & Treatments

Gorlin syndrome has many known manifestations, but the combination and degree vary from person to person. Common signs include a type of skin cancer called basal cell carcinoma (BCC), nonmalignant growths of the jaw (keratocystic odontogenic tumors or odontogenic keratocysts), small pits in the palms and soles of the feet (palmar pits), and calcification of the midline of the brain. Less common manifestations include: enlarged head, prominent forehead, skin cysts, improperly formed bones of the spine, ribs, feet and skull, fibroid tumor growth in the ovaries and/or heart, and medulloblastoma (a malignant brain tumor in young children).

Individuals with Gorlin syndrome need to see a variety of medical specialists to manage and treat their manifestations. Regular visits with a dermatologist and an oral surgeon are important. X-rays and CT scans can increase the growth of BCCs and should be limited when possible. Affected individuals should always use sunscreen and/or sun protective clothing, hats and sunglasses when outside.

Gorlin Syndrome Education

The GSA is the leading patient resource and advocacy group. Our materials are vetted by medical experts and are available to anyone impacted by Gorlin syndrome. The GSA manages a substantial library of educational resources, including a comprehensive website, physician referrals, and symptom-specific webinars.

Gorlin Syndrome Research

Gorlin syndrome research is undergoing a dramatic surge as diagnostic tools, pharmaceuticals, and biomedicine rapidly progress. The GSA serves as a liaison between these promising advancements and people with Gorlin syndrome. We manage a patient registry with anonymous data and have conducted the only global Gorlin syndrome survey. We use these tools to engage leading experts in additional research.

Gorlin Syndrome Support

We support anyone impacted by the disease, and our programs are designed to serve you. Living with a rare disease can feel lonely. Interaction with someone in your shoes can change everything. The GSA facilitates a peer-to-peer network, national/regional live and virtual meetings, and a private Facebook group. We are always available to provide support, educational materials, and a personal connection.