

What you should know about Gorlin syndrome

Individuals with Gorlin syndrome (also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS)), usually have multiple basal cell skin cancers and multiple cysts in the jaw (keratocysts), frequently starting in their teens or 20s. Most people with Gorlin syndrome develop basal cell nevi in young adulthood, but they can also occur in childhood. As they get older, individuals with Gorlin syndrome will develop more basal cell nevi. Aside from the skin cancers and jaw cysts, individuals with Gorlin syndrome may also have some characteristic physical features, including a larger than average head size, characteristic facial features, abnormalities of the ribs, spine or skull, and indentations in the palms of their hands or their feet. A person with 5 or more basal cell skin cancers or basal cell skin cancer before the age of 30 should be evaluated for Gorlin syndrome.

The risk for cancer associated with Gorlin syndrome

Individuals with Gorlin syndrome have a 90% chance to develop basal cell skin cancer. They have a 5% risk for medulloblastoma (brain cancer), which is usually diagnosed by the age of 2 years. Men and women with Gorlin syndrome also have a 2% risk for benign tumors in the heart (cardiac fibromas), and women have a 20% risk for benign tumors in the ovaries (ovarian fibromas).

The risks to family members

Gorlin syndrome is caused by mutations in the *PTCH* gene. Approximately 70-80% of individuals with Gorlin syndrome have inherited a mutation from their parents, while 20-30% have a mutation that was a new change that was not inherited. Children, brothers, sisters, and parents of a person with Gorlin syndrome all have up to a 50% chance to have the condition, and should be evaluated.

Managing the risk

Individuals with Gorlin syndrome are very sensitive to radiation. It is important that they avoid excessive sun exposure and that they not be treated with radiation therapy for cancer in order to reduce their risk for developing basal cell cancer. In individuals over age eight years, orthopantomogram every 12-18 months to identify jaw keratocysts and annual skin examinations are recommended.