UT Southwestern
Harold C. Simmons
Comprehensive Cancer Center

NCI Designated Comprehensive Cancer Center

SUFU Gene Mutations (Gorlin syndrome)

What you should know about *SUFU* mutations (Gorlin syndrome)

Gorlin syndrome (also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS)) is caused by a mutation in either the *SUFU* gene or the *PTCH1* gene. Individuals with Gorlin syndrome caused by a mutation in the *SUFU* gene usually have multiple basal cell skin cancers. As they get older, individuals with Gorlin syndrome will develop more basal cell nevi.

Aside from the skin cancers, individuals with Gorlin syndrome may also have 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. Features of Gorlin syndrome may appear more subtle or mild in individuals with a *SUFU* mutation compared to a *PTCH1* mutation. Additionally, while jaw cysts can be seen in those with *PTCH1*-related Gorlin syndrome, they have not been reported in those with *SUFU*-related Gorlin syndrome.

Cancer risks associated with a *SUFU* mutation

Individuals with Gorlin syndrome have a 90% chance to develop basal cell skin cancer. Individuals with *SUFU*-related Gorlin syndrome have up to a 33% risk for medulloblastoma (brain cancer). Males and females with Gorlin syndrome also have a 2% risk for benign tumors in the heart (cardiac fibromas). Females have a 20% risk for benign tumors in the ovaries (ovarian fibromas),.

Risks to family members

A mutation in the *SUFU* gene causes Gorlin syndrome, which is inherited in an autosomal dominant manner. This means that children, brothers, sisters, and parents of individuals with Gorlin syndrome have up to a 50% (1 in 2) chance to have Gorlin syndrome and should be evaluated. Approximately 70-80% of individuals with Gorlin syndrome have inherited a mutation from their parents, while 20-30% have a new (or "de novo") mutation that was not inherited from either parent. This new mutation can still be passed down to children.

Managing Cancer Risks

The following surveillance is recommended by the American Associated for Cancer Research (AACR), June 2017.

Annual skin examinations (with increased frequency if skin cancer observed) are recommended by age 10. Baseline echocardiogram is recommended in infancy to identify cardiac fibromas and ovarian ultrasound is recommended by age 18 to identify ovarian fibromas. Brain MRI every 4 months through age 3 and every 6 months until the age of 5 should be considered to screen for medulloblastoma.

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 skin cancer.

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