

Nevoid Basal Cell Carcinoma Syndrome [1]

What is Nevoid Basal Cell Carcinoma Syndrome?

Nevoid Basal Cell Carcinoma Syndrome (NBCCS) is also known as Gorlin syndrome. NBCCS is a hereditary condition characterized by multiple [basal cell skin cancers](#) [2] or basal cell nevus syndrome. Other common findings include jaw cysts, pits on the palms of the hands or soles of the feet, calcium deposits in the brain, and skeletal (bone) changes. The appearance of a person with NBCCS may include a larger head size, a prominent forehead, broad bridge of the nose, widely spaced eyes, skin cysts, and small skin bumps called milia. The jaw cysts and basal cell skin cancers may develop in the first ten years of a person's life, but they may not appear until the teenage years or any time during adulthood. Children with NBCCS may have the appearance features described above, including pits on their hands and feet. There is a small (5%) chance for children with NBCCS to develop a type of brain cancer called [medulloblastoma](#) [3]. Rarely, benign (not cancerous) growths in the ovaries and heart may also be found.

Multiple basal cell skin cancers and jaw cysts are the most common features of NBCCS and are present in about 90% of people who have the condition. There are several other features that have been associated with NBCCS. The number of features present and the severity of symptoms can vary among people with NBCCS, even within the same family. Black people with NBCCS may have jaw cysts as the primary feature of the disease and may develop far fewer sun-related basal cell skin cancers than people of other races with NBCCS. Jaw cysts may cause symptoms such as bone deformity, infections and pain, or they may be seen through an x-ray.

What causes NBCCS?

NBCCS is a genetic condition. This means that the cancer risk and other features of NBCCS can be passed from generation to generation in a family. The major gene associated with NBCCS is called *PTCH*. A mutation (alteration) in the *PTCH* gene gives a person an increased risk of basal cell skin cancer and other symptoms of NBCCS. Research is ongoing to learn more about NBCCS and to identify other genes involved.

Sun exposure and radiation therapy (for instance, as a possible treatment for medulloblastoma) increase the number of basal cell skin cancers that a person with NBCCS develops. Some individuals have literally thousands of basal cell cancers in areas of skin that are exposed to the sun or radiation therapy.

How is NBCCS inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. NBCCS follows an autosomal dominant inheritance pattern, in which a mutation happens in only one copy of the gene. This means that a parent with a gene mutation may pass along a copy of the normal gene or a copy of the gene with the mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a mutation also has up to a 50% chance of having the same mutation. It is also possible that the NBCCS in an individual was caused not by an inherited mutation but, rather, by a spontaneous gene mutation (see below).

How common is NBCCS?

It is estimated that about one in 40,000 people have NBCCS. As many as 30% of people with NBCCS do not have any family history of the condition. They have a *de novo* (new) mutation in the *PTCH* gene.

How is NBCCS diagnosed?

NBCCS is diagnosed when a person has at least two major features of NBCCS and one minor feature, or one major feature and at least three minor features.

Major Features:

- Multiple basal cell skin cancers [2]
- Increased calcium deposits in the head (seen on an x-ray)
- Jaw cyst(s)
- Two or more pits on the palms of the hands or soles of the feet
- A parent, sibling, or child with NBCCS

Minor Features:

- Medulloblastoma [3] in childhood
- Increased head size
- Cleft lip or palate
- Abnormal shape of the ribs or spinal bones
- Extra fingers or toes
- Eye problems
- Fibromas (benign fibrous tumors) of the ovaries or heart
- Abdominal cysts

If a person has a family history of NBCCS, that person is also suspected of having NBCCS if they have jaw cysts, multiple basal cell skin cancers, pits on the palms of the hands or soles of the feet, or calcium deposits in the head. Genetic testing for mutations in the *PTCH* gene is available for people suspected to have NBCCS. A mutation in the *PTCH* gene is found in up to 85% of families diagnosed with NBCCS.

What are the estimated cancer risks associated with NBCCS?

People with NBCCS have a 90% risk of developing multiple basal cell skin cancers [2]. About 5% of children with NBCCS will develop medulloblastoma [3].

Researchers are studying the use of medications that target the so-called "hedgehog pathway", which is affected by the *PTCH* mutation in people with NBCCS. One of these medications called vismodegib (Erivedge) has been approved to treat people with advanced basal cell cancers or those that have spread in the body. This treatment is a pill which targets the broken hedgehog gene and blocks the activated pathway. Talk with your doctor for more information about treatment options.

What are the screening options for NBCCS?

Current screening recommendations for people who are known or suspected to have NBCCS include:

- Neurologic evaluation every six months from birth to age three, then every year to age seven to look for signs of medulloblastoma [3]
- Measurement of head size regularly throughout childhood
- Yearly dental x-rays, beginning at age eight, to look for jaw cysts
- At least yearly skin exams to watch for basal cell skin cancer [2]. The frequency of exams will vary based on how many basal cell cancers or other skin problems a person has experienced. Early treatment of small basal cell skin cancer reduces the amount of surgery and scarring. Regular exams should begin by the teenage years.

Due to the high risk for multiple skin cancers, people with NBCCS should avoid sun exposure and protect their skin when outside [4]. People with NBCCS should not receive radiation therapy, as this will increase the risk of basal cell skin cancers.

Screening recommendations may change over time as new technologies are developed and more is learned about NBCCS. It is important to talk to your doctor about appropriate screening tests. In general, if there is a good screening option that doesn't use radiation, that screening option should be used to avoid skin damage in people with NBCCS.

Learn more about what to expect when having common tests, procedures, and scans [5].

Questions to ask the doctor

If you are concerned about your risk of skin cancer [2], talk to your doctor. Consider asking the following questions:

- What is my risk of skin cancer?
- What can I do to reduce my risk of skin cancer?
- What are my options for cancer screening?

If you are concerned about your family history and think you or other family members may have NBCCS, consider asking the following questions:

- Does my family history increase my risk of skin cancer?
- Should I meet with a genetic counselor?
- Should I consider genetic testing [6]?
- What preventive measures would you recommend?

Additional Resources

Guide to Skin Cancer (Non-Melanoma) [2]

Guide to Medulloblastoma, Childhood Cancer [3]

What to Expect When Meeting With a Genetic Counselor [7]

National Cancer Institute

www.cancer.gov [8]

American Cancer Society

www.cancer.org [9]

CancerCare

www.cancercare.org [10]

To find a genetic counselor in your area, ask your doctor or visit these websites:

National Society of Genetic Counselors

www.nsgc.org [11]

National Cancer Institute: Cancer Genetics Services Directory

www.cancer.gov/cancertopics/genetics/directory [12]

Links:

[1] <http://www.cancer.net/cancer-types/nevoid-basal-cell-carcinoma-syndrome>

[2] <http://www.cancer.net/node/19618>

[3] <http://www.cancer.net/node/19237>

[4] <http://www.cancer.net/node/24659>

[5] <http://www.cancer.net/node/24959>

[6] <http://www.cancer.net/node/24895>

[7] <http://www.cancer.net/node/24907>

[8] <http://www.cancer.gov/>

[9] <http://www.cancer.org>

[10] <http://www.cancercare.org>

[11] <http://www.nsgc.org/>

[12] <http://www.cancer.gov/cancertopics/genetics/directory>