CONDITION

Nevoid basal cell carcinoma syndrome

An inherited condition that increases the chance of skin cancers, jaw cysts, and certain other tumours

WHAT IT IS
Inherited cancer-predisposition condition affecting several parts of the body

BODY SYSTEM
Skin, jaw, bones, brain, and reproductive organs

MAIN SYMPTOMS
Skin cancers, jaw cysts, palm pits, skeletal differences

WHAT CAUSES IT
Inherited PTCH1 or SUFU gene change

HOW IT IS DIAGNOSED
Clinical criteria and PTCH1 or SUFU genetic testing

MAIN TREATMENT
Regular screening, skin treatment, and jaw cyst surgery

CAN IT BE CURED?
No - the inherited genetic change remains

IS IT AN EMERGENCY?
Sometimes - new seizures, weakness, or severe sudden headache

LONG-TERM OUTLOOK
Often manageable, but repeated tumours and cysts are common

Overview

Nevoid basal cell carcinoma syndrome is an inherited condition that affects how cells grow and develop. It increases the chance of developing basal cell carcinomas, a common type of skin cancer. In this condition, these cancers may appear at a younger age and in larger numbers than usual.

The condition can also cause cysts in the jaw, small pits on the palms or soles, and differences in the bones or shape of the head. Some people develop other benign tumours, meaning growths that are not cancer. There is also an increased risk of certain cancers, including a childhood brain tumour called medulloblastoma.

The effects differ greatly between people. Some have many skin cancers and need repeated treatment. Others have fewer visible signs and learn they have the condition only after a jaw

cyst, genetic test, or diagnosis in a relative.

It matters because regular checks can find many complications early, when they are easier to manage. Care often involves dermatology, dental or jaw specialists, genetics services, and sometimes other teams.

Symptoms

Features may begin in childhood, adolescence, or adulthood. They may not all appear at the same time.

Basal cell carcinomas often look like pearly, pink, brown, or skin-coloured bumps. They may form a sore that does not heal, bleed repeatedly, crust over, or slowly enlarge. Although they rarely spread to distant organs, untreated lesions can damage nearby skin, cartilage, bone, or nerves.

Jaw cysts, called odontogenic keratocysts, usually develop inside the jawbone. They may cause swelling, pain, tooth movement, or problems with tooth eruption. Some are found during dental imaging before they cause symptoms.

Other possible features include:

Medulloblastoma is uncommon but is an important risk, especially in young children with certain SUFU gene changes. Possible symptoms include persistent or worsening headaches, repeated vomiting, poor balance, unusual clumsiness, or changes in behaviour or alertness.

Having one or two of these features does not by itself mean that you have this condition.

- Tiny pits on the palms of the hands or soles of the feet
- A larger-than-average head or a prominent forehead
- Rib, spine, skull, or other skeletal differences
- Small calcium deposits inside the skull that usually cause no symptoms
- Benign fibromas, which are firm growths, in the ovaries or heart
- Eye differences, such as crossed eyes or unusually wide-set eyes
- Developmental or learning difficulties in some people

Causes and risk factors

Nevoid basal cell carcinoma syndrome is usually caused by a harmful change in the PTCH1 or SUFU gene. These genes help control a cell-signalling system called the hedgehog pathway.

This pathway guides normal growth before birth and helps regulate cell growth later in life. A harmful gene change can allow some cells to grow when they should not.

The condition follows an autosomal dominant inheritance pattern. This means a person with the gene change has a 1 in 2 chance of passing it on during each pregnancy. The chance is the same for children of any sex.

Some people inherit the change from a parent. In others, it begins as a new genetic change, so there may be no earlier family history. Occasionally, the change is present in only some of the body's cells. This is called mosaicism and can make genetic testing and diagnosis more complicated.

Ultraviolet radiation from sunlight or tanning devices does not cause the inherited condition. However, it can further increase the number of basal cell carcinomas that develop.

Radiation used for cancer treatment can also trigger numerous basal cell carcinomas in the treated area. Medical imaging uses much lower doses, but clinicians usually try to avoid unnecessary exposure to ionising radiation, such as repeated X-rays or CT scans. Imaging that is medically necessary should not be delayed without discussing the risks and alternatives with the relevant team.

Diagnosis

Diagnosis may be considered when a person has a characteristic combination of basal cell carcinomas, jaw cysts, palm or sole pits, skeletal findings, or an affected close relative.

A clinical genetics service can review your medical history, family history, skin findings, dental records, and imaging. There are established clinical criteria that combine major and minor features. No single visible feature confirms the condition.

A blood or saliva test may look for harmful changes in PTCH1 and SUFU . Finding a disease-causing change can confirm the diagnosis and help guide screening. It can also allow relatives to have targeted testing for the same family change.

A negative genetic test does not always rule the condition out. Current testing may not find every possible gene change, and mosaic changes can be harder to detect. Clinical findings may therefore remain important.

A skin biopsy is used to confirm whether an individual growth is a basal cell carcinoma. During a biopsy, a small tissue sample is removed and examined under a microscope. Jaw cysts are also examined after removal to confirm their type.

Further assessment is based on age, symptoms, gene result, and previous findings. It may

include skin examinations, dental or jaw imaging, and brain MRI in children at increased risk of medulloblastoma. MRI does not use ionising radiation.

Treatment and management

Treatment cannot remove the inherited gene change. The aim is to find and treat tumours or cysts early, limit tissue damage, and reduce the effects of repeated procedures.

Basal cell carcinomas may be removed surgically. The exact technique depends on their size, depth, location, and number. Some superficial lesions can be treated with prescription creams, scraping and cautery, freezing, light-based treatment, or other local procedures.

When there are many basal cell carcinomas, care may need to balance cancer control with scarring and the burden of repeated treatment. Medicines called hedgehog pathway inhibitors may be considered for selected adults with numerous, advanced, or difficult-to-treat cancers. These medicines can have significant side effects, and tumours may return after treatment stops. They require specialist supervision.

Radiotherapy is generally avoided when another suitable treatment is available because it can cause many new basal cell carcinomas in the exposed area. There are situations in which radiotherapy may still be considered after a careful discussion of benefits and risks.

Jaw cysts usually require treatment by an oral and maxillofacial surgeon. Options include removing the cyst or first opening it to reduce its size. Cysts can return, so follow-up imaging may continue after treatment.

Other care is tailored to the findings. An ovarian fibroma may need gynaecological assessment if it causes pain, bleeding, pressure, or concern about twisting of the ovary. A heart fibroma may be monitored or treated depending on its size, location, and effect on heart rhythm or function.

Genetic counselling can help you understand test results, family testing, and reproductive options without directing you toward a particular choice.

Complications

The most common long-term problem is the repeated development of basal cell carcinomas. These cancers usually grow locally rather than spreading through the body. Even so, they can damage nearby tissue if treatment is delayed, particularly around the eyes, nose, ears, or lips.

Jaw cysts can become infected, weaken the jawbone, displace teeth, or return after removal.

Repeated surgery may affect teeth, nerves, or the shape and strength of the jaw.

Other possible complications include:

Not everyone develops these complications. Risk patterns can differ between PTCH1 -related and SUFU -related forms of the condition.

- Scarring or changes in appearance after repeated skin procedures
- Pain, infection, or numbness related to jaw cysts and surgery
- Medulloblastoma, mainly during early childhood
- Ovarian twisting or pressure from an ovarian fibroma
- Heart rhythm or blood-flow problems from a cardiac fibroma
- Emotional strain related to screening, procedures, and visible skin changes

Prevention

The inherited condition itself cannot currently be prevented. A person does not cause it through diet, lifestyle, or anything done during pregnancy.

Sun protection cannot remove the genetic risk, but it can reduce additional ultraviolet damage that may contribute to basal cell carcinomas. Useful measures include shade, protective clothing, a wide-brimmed hat, and broad-spectrum sunscreen. Avoid tanning beds and deliberate tanning.

Check your skin regularly so you become familiar with existing marks and scars. Photographing an area can help you notice change over time, although photographs do not replace a professional skin examination.

Tell dental, medical, and imaging teams about the condition. This can help them avoid duplicate or unnecessary X-rays and consider MRI or ultrasound when those tests can answer the same question. Necessary imaging should still be completed when its expected benefit is greater than the radiation risk.

If a family gene change is known, genetic counselling can explain testing before or during pregnancy, including preimplantation genetic testing. These are personal options rather than required preventive steps.

Living with it and what to expect

Nevoid basal cell carcinoma syndrome is lifelong. Bone differences and palm or sole pits

usually remain, but may not need treatment. The main changes over time are the possible appearance of new skin cancers and the development or recurrence of jaw cysts.

Skin surveillance continues through adult life because basal cell carcinomas may keep appearing. The number and timing are difficult to predict. Some people need only occasional procedures, while others require regular treatment over many years.

Jaw cysts often begin during childhood or adolescence, particularly with PTCH1 changes. Follow-up may continue after removal because recurrence is possible. Medulloblastoma risk is concentrated in childhood, so brain surveillance, when needed, is planned differently from lifelong skin checks.

Most people can attend school, work, exercise, form relationships, and have children. Appointments and repeated procedures can still disrupt daily life. Visible scars or worry about cancer may affect confidence or mood. Psychological support, practical planning around appointments, and patient support groups can be useful.

Keep a record of genetic results, pathology reports, operations, and recent imaging. Because several specialties may be involved, this record can reduce repeated testing and help new clinicians understand your history.

Relatives may have much milder or more noticeable features than you, even when they carry the same gene change. Your own course cannot be predicted from one relative's experience.

When to get medical help

Arrange routine medical or dermatology review if you notice:

These concerns usually need timely assessment but are not emergencies.

Seek urgent medical assessment for repeated vomiting, a worsening headache, new balance problems, marked drowsiness, or a sudden change in behaviour, especially in a child known to be at increased risk of medulloblastoma.

Call emergency services for a new seizure, sudden weakness, loss of consciousness, severe breathing difficulty, or a severe sudden headache. Sudden intense pelvic pain with nausea or vomiting also needs emergency assessment because an ovarian growth can sometimes twist and interrupt its blood supply.

- A new or changing skin growth
- A sore that repeatedly bleeds, crusts, or does not heal
- New jaw swelling, dental movement, or persistent jaw discomfort

- Increasing pain or changes near a previous cyst or operation
- New pelvic pressure, swelling, or persistent pain
- Concerns about screening, family testing, or treatment side effects

Common questions

Is nevoid basal cell carcinoma syndrome itself cancer?

No. It is an inherited condition that raises the chance of certain cancers and benign tumours. Each suspicious skin growth or other mass needs its own assessment. Many findings linked to the condition are not cancer.

Will I definitely develop basal cell carcinomas?

Not everyone has the same skin findings or tumour burden. Your risk is influenced by the gene involved, age, sun exposure, previous radiation, and other factors that are not fully understood. Regular skin examinations are important even if you have never had skin cancer.

Can I pass it to my children?

If you have a harmful PTCH1 or SUFU change in all your cells, each child has a 1 in 2 chance of inheriting it. Mosaicism can change this estimate. A genetics service can explain what your specific test result means.

Why are X-rays sometimes limited?

Ionising radiation can add to the risk of new tumours, so unnecessary or repeated exposure is avoided where practical. This does not mean all X-rays or CT scans are unsafe. A medically necessary scan may provide much more benefit than risk.

Do all skin growths need surgery?

No. Treatment depends on whether the growth is cancer, its size and location, and how many lesions are present. Surgery is common, but selected superficial lesions may have other local treatments. Some harmless marks only need monitoring.

Can I have normal dental treatment?

Usually, yes. Routine dental care remains important. Tell your dentist about the condition and any history of jaw cysts. Dental and jaw teams can plan appropriate imaging, watch for cysts, and avoid unnecessary repeat examinations.

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