



NEVOID BASAL CELL CARCINOMA SYNDROME

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Nevoid Basal Cell Carcinoma Syndrome (NBCCS) is also known as **Basal Cell Nevus Syndrome** and **Gorlin Syndrome**. A dentist named R. J. Gorlin first identified the syndrome in 1960, but Egyptian mummies 4000 years old have been found with evidence of the disease. NBCCS is caused by a genetic mutation usually inherited from a parent, however up to one third of cases occur in the absence of any family history. It is a rare disorder, affecting between 1/56,000 to 1/164,000 people of all races with equal frequency in men and women.

The most identifiable feature of NBCCS is multiple **basal cell carcinomas**, a common type of skin cancer. This type of skin cancer is most often seen in older adults after many years of sun damage. People with NBCCS frequently get basal cell carcinomas in their 20s and 30s, though can be as young as 3-4 years old. They also have many more basal cell cancers; the average number for people with the syndrome is 8, but they can have thousands. Skin type and exposure to sun and radiation damage strongly influence the number of basal cell cancers patients with the syndrome develop. Black individuals with NBCCS typically develop relatively few skin cancers, whereas fair-skinned white people with the syndrome can have many.

In addition to skin cancer, people with the syndrome often have pits, or small indentations, in the skin of their palms or soles that appear over time and are permanent. Multiple **cysts** (milia, epidermal cysts) and **moles** or **skin tags** are seen in people with NBCCS. The syndrome also affects the bones, face, and brain. Many of the most common characteristics of NBCCS do not cause symptoms.

Jawbone cysts (odontogenic keratocysts) can be the first sign of NBCCS and are often found by a dentist. The cysts first appear in children and continue to form until approximately 30 years of age. Sometimes they are asymptomatic, but they can be painful, displace teeth, and cause swelling. Other skeletal abnormalities include cysts in the hand or foot bones, deformed ribs or spinal vertebrae, scoliosis, spina bifida, shortened fourth and fifth fingers, fused fingers (syndactyly), and an abnormality of the shoulder blade called Sprengel deformity.

Atypical facial features can be seen in NBCCS. Up to 70% of patients have a broadened face, coarse facial features, or wide-set eyes (ocular hypertelorism). Enlarged head circumference (macrocephaly) and cleft lip or cleft palate, are other findings. People with NBCCS have a higher incidence of cataracts at birth, crossed eyes (strabismus), and bulging of the eyes (exophthalmos).

Abnormal areas of calcification in the brain are seen in many people with NBCCS. The areas of calcification (falx cerebri, tentorium cerebelli, interclinoid ligament of the sella turcica) can be identified on x-ray of the skull. There can be cysts in the brain and sometimes the bundle of nerve fibers that connect the two hemispheres of the brain fails to develop (agenesis of the corpus callosum). Mental retardation occurs in approximately 5% of cases. Brain cancers, specifically medulloblastoma and meningioma, occur in 1-4% of children with the NBCCS.

People with NBCCS are at higher risk for some other cancers and benign growths than the general population. A type of bone cancer (fibrosarcoma) and muscle cancer (rhabdomyosarcoma), while not common, do occur slightly more often in NBCCS. A non-cancerous growth in the heart (cardiac fibroma) is sometimes seen in children with this syndrome. If present, this growth can be life-threatening depending on its size and location in the heart. Ovarian cysts and fibromas are found in approximately 25% of cases. These benign growths do not affect fertility.

The signs and symptoms of the syndrome identify NBCCS. A diagnosis is made by the presence of two major criteria or one major and two minor criteria.

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Major criteria:

- More than 2 BCCs or 1 BCC diagnosed before 20 years of age
- Jaw cysts
- Three or more skin pits on the palms or soles
- Calcification of falx cerebri in the brain
- Deformities of the ribs
- First degree relative with NBCCS

Minor criteria:

- Enlarged head circumference
- Congenital malformation (cleft lip or palate, coarse facial features, wide-set eyes)
- Structural anomalies (Sprengel deformity, syndactyly)
- Abnormalities on x-ray (spine malformation, cysts in bones of hands or feet)
- Ovarian fibromas
- Brain cancer (medulloblastoma)

X-ray, ultrasound, and MRI are needed to recognize many of these criteria. Genetic testing is not done in most cases because it is not very accurate. Genetic testing may be indicated in infants of parents with the syndrome for early identification purposes or in cases that are unusual and difficult to diagnose.

One of the most important aspects of treating NBCCS is early identification and removal of basal cell carcinoma. Patients should see a dermatologist at least every three months for screening. Early detection is critical so that skin cancers can be taken care of while they are small in the least invasive manner possible. Metastasis of basal cell carcinoma is rare, but if not removed these cancers can be locally destructive. Tumors can be treated with topical therapy (**fluorouracil** or **imiquimod**), burning and scraping of the lesion (**electrodessication and curettage**), excision, or **Mohs micrographic surgery**.

Patients with NBCCS need to be very careful to avoid sun exposure. Sun protective clothing, hats, and **sunscreen** should always be used when outside. Radiation therapy should be avoided, as this also increases skin damage and the development of basal cell cancers.

Other medical care may be indicated depending on an individual's risk factors and complications. Neurology, cardiology, gynecology, oral surgery, and ophthalmology may be involved with screening and care of patients with NBCCS. Because NBCCS is an inherited condition, patients with the syndrome who are considering having children should seek genetic counseling.

This syndrome can be emotionally difficult for patients. Frequent doctor visits, recurrent cancers and surgeries, and the resulting scars can take a toll on patients. It is important to address this burden. Patients can find support at the Basal Cell Carcinoma Nevus Syndrome Life Network (gorlinsyndrome.org).

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