Carney Complex (CNC)

Carney complex (CNC) is a rare genetic condition in which people typically have pigmented (dark) areas on their skin, as well as a higher risk of certain types of tumors.

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What are the effects of Carney complex (CNC)?

In addition to having dark, pigmented areas on their skin, people with CNC are at higher risk for certain tumors including:

- **Endocrine tumors:** Tumors in organs or glands that make hormones, such as the pancreas, testicles, ovaries, or the adrenal, thyroid, parathyroid, or pituitary glands
- **Myxomas:** Benign (non-cancerous) tumors that develop most often in the heart (cardiac myxomas) but can also start in the skin or other organs
- **Schwannomas:** Tumors (usually non-cancerous) that start in Schwann cells, which normally wrap around and insulate nerves

Signs of CNC typically appear during childhood or when a person is in their teens or early 20s.

Hyperpigmented (darker) skin spots are the most common sign of CNC. They tend to
increase in number during puberty and may fade as people get older.

Cardiac myxomas are also common. While these tumors are non-cancerous (because they don’t spread to other parts of the body), they can still cause serious health issues. These tumors can reduce blood flow in the heart and increase the risk of blood clots (which can lead to a stroke), and they can be life-threatening. Myxomas can also develop on the skin or in the mouth, breast, or other organs.

Endocrine tumors of the adrenal and pituitary glands and of the testicles (in males) are also common in people with CNC. Tumors in these glands or organs can result in abnormally high levels of specific hormones in the body:

- In adrenal gland tumors, high levels of cortisol can lead to weight gain, high blood pressure, diabetes, and easy bruising. This is known as primary pigmented nodular adrenocortical disease (PPNAD).
- Pituitary gland tumors can lead to high levels of growth hormone, but this (known as asymptomatic growth hormone hypersecretion) usually doesn’t cause any symptoms. Some people may develop acromegaly (enlarged facial features, hands, and feet).
- In males with Sertoli cell tumors in the testicles (known as large cell calcifying Sertoli cell tumors, or LCCSTs), high levels of estrogen can cause enlargement of breast tissue, short height, and early puberty.

What causes Carney complex (CNC)?

Most often, CNC is caused by a mutation in the PRKAR1A gene. More than 130 different PRKAR1A mutations have been found. Most of these mutations have only been identified in a single family.

Changes in other genes are also thought to cause CNC in some people.

Most people with CNC inherit a mutated PRKAR1A gene from a parent. A person with CNC has a 50/50 chance of passing the PRKAR1A gene mutation on to each of their children. However, in about 1 in 4 people with CNC, the gene mutation isn’t inherited from a parent. Instead, the mutation is a random event that happens very early in a person’s life (possibly even before birth). Then it can be passed down to that person’s children.

Options exist for people who carry a PRKAR1A gene mutation and might want to have
children. For more information, talk with an assisted reproduction specialist at a fertility clinic.

**How common is Carney complex (CNC)?**

CNC is rare. Only several hundred cases have ever been reported worldwide.

**How is Carney complex (CNC) diagnosed?**

People are diagnosed with Carney complex based mainly on if they have certain criteria. CNC can be diagnosed:

- If you have 2 or more of the major diagnostic criteria below, OR
- If you are found to have a mutation in the *PRKAR1A* gene, OR
- If you have 1 major diagnostic criterion and a first-degree family member (parent, sibling, or child) with CNC

**Major diagnostic criteria for CNC include:**

- Spotty skin pigmentation
- Myxoma (of the heart, breast, skin, or mouth or other mucosal surface)
- Breast ductal adenoma (benign breast tumor)
- Primary pigmented nodular adrenocortical disease (PPNAD)
- Acromegaly (enlarged facial features, hands, and feet due to excess growth hormone from a pituitary tumor)
- Large cell calcifying Sertoli cell tumor (LCCST)
- Thyroid cancer\(^1\) or multiple thyroid nodules in young person
- Schwannoma (tumor of the nerve sheath)
- Blue nevi (blue-black moles)
- Osteochondromyxoma (a type of bone tumor\(^2\))

**How often should people with Carney complex (CNC) be screened for tumors?**

Because Carney complex is so rare, there are no standard screening guidelines for people with CNC. However, recommendations from medical experts to look for tumors early typically include:
A yearly **echocardiogram** (ultrasound of the heart), beginning in infancy. People who have already had a cardiac myxoma should have an echocardiogram every 6 months.

- **Yearly endocrine (hormone) blood tests** such as cortisol, prolactin, and IGF-1, beginning in a person’s teens.
- **Yearly thyroid ultrasound.**
- **Yearly testicular ultrasound** for boys, starting before puberty.

Screening recommendations may change over time as new technologies are developed and more is learned about Carney complex. It’s important to talk with your health care team about appropriate screening tests.

**How is Carney complex (CNC) managed?**

Treatment of CNC is different for each person, depending on the type of tumor(s) they have, their symptoms, and other factors.

In general, most types of tumors resulting from Carney complex are removed with surgery if they are causing symptoms (or if they might cause problems in the future). Other types of treatments might also be helpful in some situations. Close follow-up is usually recommended after treatment to watch for possible signs of a tumor recurring (coming back) or new tumors forming.

For some types of tumors that aren’t likely to cause health issues, watching them closely (instead of removing them right away) might be an option.

**Questions to ask the health care team**

If you are concerned about your risk of Carney complex or the tumors it can cause, talk with your health care team. It might be helpful to bring someone along to your appointments to take notes. Consider asking your health care team the following questions:

- Could my family carry a gene mutation for Carney complex? How likely is it that I might have it?
- Should I meet with a genetic counselor or other genetics specialist for a risk assessment, and possibly **genetic testing**? Can you refer me to someone?
- Should I get any screening tests to look for tumors? When should these start?
Hyperlinks


References


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