Cowden Syndrome

Cells are the basic units of life and are the building blocks for every part of the body. Genes are pieces of information within cells. Genes tell the cells in the body what to do.

Genetic conditions are caused by changes (mutations) in a person’s genes. A gene mutation can cause the gene to stop working correctly and can cause medical problems.

Cowden syndrome (CS) is a genetic condition caused by a mutation in the PTEN gene. The PTEN gene normally works to protect the body from forming certain types of tumors. When the PTEN gene stops working normally, the risk of developing these tumors increases.

People with CS have an increased risk of developing certain benign (non-cancerous) and cancerous tumors.

- People with CS have an increased risk of developing thyroid cancer.
  - The thyroid is a gland in the base of the throat that helps make hormones.
  - There are different types of thyroid cancer. Follicular thyroid cancer is the most common type that occurs with CS. Medullary thyroid cancer not related to CS.
- Women with CS have an increased risk of developing breast and uterine cancer.
- People with CS have an increased risk of developing certain types of colon polyps.
- Many different benign tumors are also common in people with CS.

CS is a rare condition. Researchers are still learning about the cancer risks caused by CS. **If you have CS, it is important that you stay in contact with your genetics clinic.** Your care team will help you learn about new research findings and how it applies to you.

Cowden syndrome is sometimes called PTEN hamartoma tumor syndrome (PHTS). There is also a rare type of CS called Bannayan-Ruvalcaba-Riley syndrome.

Signs and Symptoms

CS affects each patient differently. There are many possible signs and symptoms of CS. It is most common to have some (not all) of these symptoms:

- Learning disabilities, autism and/or an intellectual disability
- Large head size
- Certain types of lesions or papules (bumps) on the skin. A dermatologist can recognize the most common, including:
  - Trichilemmomas on the face
  - Papillomatous lesions, particularly if they are on the face and/or mucous membranes (such as gums). A “cobblestone” appearance on the tongue or gums may happen.
  - Keratoses (hard growths on the skin) found on the palms of the hands or soles of the feet
Cancer Risks

Women

- Have up to an 85% chance of developing breast cancer throughout their life. This is higher than the average woman’s chance of developing breast cancer (about 12%). Women with CS are also more likely to develop breast cancer at a younger age (38-50 years of age).
- Have up to a 28% chance of developing endometrial (uterine) cancer throughout their lifetime. This is higher than the average woman’s chance of developing endometrial cancer (3%).

Men and Women

- Have a 10 to 35% chance of developing thyroid cancer. This is higher than the average man and woman’s risk (of 1%).
- May be at increased risk for other types of cancers, such as kidney and colon cancer. The specific risk for these types of cancers is unknown at this time.
- Rarely, adults with CS develop a type of brain tumor called Lhermitte-Duclos disease.

Benign Tumor Risks

People with CS may have an increased chance to develop the following benign tumors. These tumors are also common in the general population.

- A goiter (enlarged thyroid) or a benign tumor in the thyroid
- Polyps in the stomach, small intestine or colon
- Uterine fibroids (growth in the uterus)
- Fibrocystic breast changes (a feeling of lumps and/or tenderness in the breasts)
- Lipomas (benign fatty tumors) and fibromas (benign tumors of connective tissue)

Diagnosis

Diagnosing CS can help plan for additional cancer screenings and preventive care. This can help to detect cancer at an earlier stage. Even if a person with CS already has cancer, there is a risk that a second cancer may develop.

Most often, diagnosing CS starts with 2 evaluations:
1. A physical exam – including a skin check by a dermatologist who is familiar with CS.
2. A review of family history – a genetic counselor reviews detailed family history in order to look for signs of CS in the family.

Genetic Testing

If the evaluations show signs of CS, PTEN genetic testing is most often the next step. Genetic testing uses a blood or saliva sample to look for mutations in the PTEN gene.

- If the test finds a PTEN mutation, the patient is diagnosed with CS.
- Genetic tests are not perfect. Even if the test does not find a PTEN mutation, the patient may have CS. A doctor who is familiar with genetics or a genetic counselor can best understand and explain the test results.
Family Members

CS is a genetic condition, so family members are also at risk. The parents of a person with CS may or may not have CS.

- It is possible for a person to be born with CS and be the first person in the family with the PTEN mutation (this happens in more than half of cases).
- A person with CS has a 50% (1 in 2) chance to pass the PTEN gene mutation to each child.
- Men and women have equal chance of having CS.
- CS does not skip generations.

Family members may benefit from genetic testing when a person in the family has CS. Genetic testing can help to find out if other family members have CS. This can help doctors decide which family members should consider additional cancer screenings and preventive care.

Screening and Prevention

Screening helps detect cancer as early as possible when it may be easier to treat. There is no cure for CS. Because cancer is the major health risk related to CS, it is important to follow cancer prevention and early detection screening guidelines.

**Breast Cancer Screening (Women)**

- At age 18, start monthly self-breast exams (patient checks herself).
- At age 25, have a breast exam by a medical provider every 6 months.
- At age 30 to 35, have a mammogram and breast MRI (imaging tests) every year.
- Women with CS may consider having preventive surgery (removing the breasts before cancer develops).

**Thyroid Cancer Screening (Men and Women)**

- At age 7, have a thyroid ultrasound (imaging test) every year.
- If diagnosed with CS after age 7, start yearly thyroid cancer screenings when diagnosed.

**Other Screenings**

- At age 18, have a complete physical exam every year.
- Consider a skin exam by a dermatologist every year.
- At age 35, have a colonoscopy every 5 years. This is an exam of the inner lining of the large intestine (colon and rectum). Your doctor may recommend frequent colonoscopies if polyps are found.
- At age 40, consider a kidney ultrasound (imaging test) every 1 to 2 years.
- Women with CS should talk with a gynecologist or gynecologic oncologist about their uterine cancer risk. A doctor can explain options for screening or preventive surgery (removing the uterus before cancer develops).
- When children are diagnosed with CS, consider cognitive and motor assessments.
More Resources

Genetic Alliance
202-966-5557
http://www.geneticalliance.org
This organization provides support to individuals and families with genetic conditions. A helpline is available to answer questions.

American Cancer Society
800-ACS-2345 (800-227-2345)
www.cancer.org
The American Cancer Society (ACS) is a voluntary national health organization with local offices around the country. The ACS supports research, provides information about cancer, and offers many programs and services to patients and their families.

Cancer Information Service
800-4-CANCER (800-422-6237)
www.cancer.gov
The Cancer Information Service (CIS) is a program of the National Cancer Institute. Highly trained information specialists are available to answer questions about cancer screening tests, risks, symptoms, how cancer is diagnosed and the newest treatments.

Susan G. Komen Breast Cancer Foundation
800-462-9273
www.breastcancerinfo.com
The Komen Foundation answers questions from recently diagnosed breast cancer patients and provides emotional support. Information is available in Spanish.

National Society of Genetic Counselors, Inc. (NSGC)
312-321-6834
FYI@nsgc.org
www.nsgc.org
This professional organization helps consumers find local genetic counseling services.