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 in a person's genes, also called a mutation. A gene mutation can cause the gene to stop working correctly and cause medical problems.

Cowden syndrome (CS) is a genetic condition caused by a mutation in the *PTEN* gene. The *PTEN* gene normally protects the body from forming certain types of tumors. A mutation in the *PTEN* gene increases the risk of developing these tumors.

CS is a rare condition and there is no cure. 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 they apply to you.

Signs and Symptoms

Cowden syndrome affects each patient differently. There are many possible signs and symptoms of CS. It is common to have some, but not all, of these symptoms:

- Learning disabilities, autism or an intellectual disability
- Large head size
- Certain types of lesions or bumps on the skin. A dermatologist can recognize the most common, including:
  - Benign, meaning non-cancerous, tumor on hair follicles called trichilemmomas. These are found on the face.
  - Benign, wart-like growths called papillomas, particularly if they are on the face or mucous membranes, such as gums. The tongue or gums may have a "cobblestone" appearance.
  - Hard growths on the skin, called keratoses, on the palms of the hands or soles of the feet.

Cancer Risks

People with Cowden syndrome have an increased risk of developing certain types of cancer.

Type of Cancer	How many people with CS may develop this cancer	How many people in the general population may develop this cancer
Breast cancer	60 people out of 100 people	12 people out of 100 people

Uterine cancer	28 out of 100 people	3 out of 100 people
Kidney cancer • Papillary renal cell carcinoma is the most common type with CS.	35 people out of 100 people	2 people out of 100 people
Thyroid cancer • Follicular thyroid cancer is the most common type with CS.	10 to 35 out of 100	1 out of 100
Colon cancer	9 out of 100	5 out of 100
Melanoma	6 out of 100	3 out of 100

Rarely, adults with CS can develop a type of brain tumor called Lhermitte-Duclos disease.

Benign Tumor Risks

People with CS may have an increased chance to develop benign, non-cancerous tumors:
- An enlarged thyroid, also called a goiter, or a benign tumor in the thyroid
- Polyps in the stomach, small intestine or colon
- Growths in the uterus, also called uterine fibroids
- A feeling of lumps or tenderness in the breasts
- Benign fatty tumors, also called lipomas
- Benign tumors of connective tissue, also called fibromas

Diagnosis

Diagnosing Cowden syndrome can help plan for more cancer screenings and preventive care. This can help detect cancer at an earlier stage. There is a risk that a second cancer may develop, even if you currently have cancer.

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

Genetic Testing

If the evaluations show signs of CS, genetic testing for the *PTEN* gene is often the next step. Genetic testing uses your blood or saliva to look for mutations in the *PTEN* gene.
- If the test finds a *PTEN* mutation, you will be diagnosed with CS.
- Genetic tests are not perfect. Even if the test does not find a *PTEN* mutation, you may still have CS. A doctor who is familiar with genetics or a genetic counselor can best understand and explain the test results.

Family Members

Cowden syndrome 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 1 in 2 chance to pass the *PTEN* gene mutation to each of their children.
- Males and females 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 find out if other family members have CS. This can help doctors decide which family members should consider more cancer screenings and preventive care.

Screening and Prevention

Screening helps detect cancer as early as possible when it may be easier to treat. It is important to follow cancer prevention and early detection screening guidelines.

Breast Cancer Screening

- At age 18, start monthly self-breast exams.
- At age 25, start breast exam completed by a medical provider every 6 months.
- At age 30, start mammograms and breast magnetic resonance imaging (MRI) every year.
- You may consider having preventive surgery, such as removing the breasts before cancer develops. Age to start breast screening may change based on family history of breast cancer.

Thyroid Cancer Screening

- At age 7, start thyroid 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.
- A skin exam by a dermatologist every year, starting now.
- At age 35, have a colonoscopy every 5 years. This is an exam of the inner lining of the colon and rectum. Your doctor may recommend more frequent colonoscopies if polyps are found.
- At age 40, consider a kidney ultrasound imaging test every 1 to 2 years.
- Talk with your gynecologist or gynecologic oncologist about your uterine cancer risk. A doctor can explain options for screening or preventive surgery, such as removing the uterus before cancer develops. Uterine cancer screening could be considered at age 35.
- If your child is diagnosed with CS, consider cognitive and motor assessments.

Resources

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

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

American Cancer Society
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.