Cancer Genetics Summary

Cowden Syndrome (Gingival Multiple Hamartoma Syndrome)

Cowden Syndrome is a rare disorder characterized by multiple noncancerous, tumor-like growths called hamartomas and an increased risk of developing certain cancers. In addition to harmatomas, in affected individuals the syndrome can cause:

- Large head size (50 percent likelihood)
- Abnormal breast findings (75 percent of affected females)
- Thyroid conditions (67 percent)
- Gastrointestinal polyps (60 percent)
- Genitourinary malformations (40 percent)

Approximately 1 in 200,000 people are diagnosed with Cowden Syndrome. The actual incidence is likely to be higher because the syndrome is difficult to diagnose.

Cowden Syndrome has been linked to a mutation in the PTEN gene. About 30 percent of PTEN mutations are inherited; the remainder result from new mutations. A mutation in this gene will result in an increased risk for breast, thyroid and uterine cancers. In addition, several studies have reported increased risks for melanoma, glioblastoma, certain types of kidney cancer and transitional cell carcinoma of the bladder.

PTEN Cancer Risks

Breast cancer is the most common cancer in people carrying an altered PTEN gene. Women who carry a PTEN mutation have a 20 to 50 percent risk of developing breast cancer, compared to 11 percent in the general population. There is also a 35 percent chance that if cancer occurs, both breasts will be affected. The average age of onset of breast cancer in Cowden Syndrome is 38 but it has been seen as young as age 14.

People affected with Cowden Syndrome have a 10 percent risk for thyroid cancer, compared to 1 percent in the general population. The risk for uterine cancer, uterine fibroids and uterine polyps are also increased.

PTEN Gene

The tumor suppressor gene implicated in the majority of Cowden Syndrome families is PTEN (Phosphatase and Tensin homolog deleted on chromosome Ten), located on the long arm of chromosome 10. This gene is responsible for inhibiting uncontrolled cell growth. When the gene is mutated, the cells may no longer be able to regulate cell growth and this can lead to tumor formation.

Genetic Testing for Cowden Syndrome

Gene testing is available for mutation analysis of the PTEN gene. A mutation in the PTEN gene will confirm a diagnosis of Cowden Syndrome and the risks associated with the syndrome. Predisposition gene testing is best performed on someone who has had cancer. If that individual is found to have a change in the suspect gene, then other relatives can be reliably tested.

If a mutation in the PTEN gene is found in your family but your test results show that you did not inherit the gene mutation, your chance of developing cancer would be the same as for the general population. If you do not have the gene mutation, your children will not inherit the mutation from you.

For more information or questions, call 503-413-6534.

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