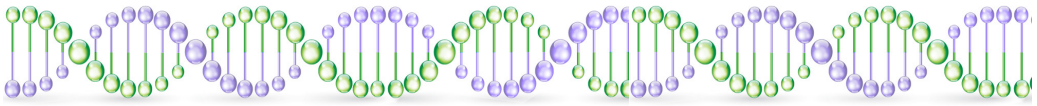
Information Sheet



SYDNEY CANCER GENETICS

Cowden syndrome, PTEN Hamartoma Tumour syndromes and the PTEN gene

When there is a mutation (change in the DNA code) in the PTEN gene, the signs and symptoms that occur can vary greatly, even from individual to individual in the same family. PTEN mutations are associated with 3 different syndromes:

- Cowden syndrome (discussed below)
- Bannayan-Riley-Ruvalcaba syndrome (associated with a large head (macrocephaly), hamartomatous polyps in the intestine, lipomas and freckles on the penis)
- Proteus and Proteus-like syndromes ("overgrowth" syndromes, usually diagnosed in childhood).

What is Cowden syndrome?
Cowden syndrome, also known as a PTEN Hamartoma syndrome, is a heritable cancer syndrome that affects 1:200,000 people.

Characteristic skin lesions develop over time and are present in almost all people with Cowden syndrome by the late 20s. These lesions include trichilemmomas, papillomatous papules, acral keratosis and lipomas. You can see some examples on our website. They are not cancerous and usually require no treatment.

Macrocephaly (head circumference on the 97th percentile) is common and, in some individuals, intellectual delay.

Benign growths, such as hamartomas, are very common. These growths occur in the breast (benign fibrocystic breast disease in 80%), the thyroid (multinodular goitre in 60%), uterus (fibroids in 40%) and bowel (hamartomatous polyps in 30%).

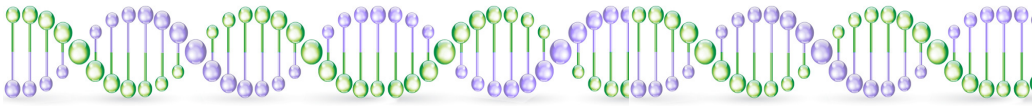
Does Cowden syndrome cause cancer?
The risk of cancer is increased in individuals with a PTEN mutation:

- the risk of breast cancer for women is 25% over a lifetime
- the risk of uterine cancer for women is 5 to 10% over a lifetime
- the risk of thyroid cancer is 5 to 10% over a lifetime.
- the risk of kidney cancer is also 5 to 10% over a lifetime.

How is Cowden syndrome managed?
To reduce cancer risk and/or detected cancers early:

- breast cancer screening for women should start at age 30
- a hysterectomy should be considered at age 40 (after the woman has completed her family)
- an ultrasound of the kidneys should be considered every 2 years from age 40
- bowel cancer screening with colonoscopies should be considered every 5 years from age 35 or 40
- to check the thyroid, a clinical examination of the neck by the GP is recommended every year from age 5. Ultrasounds of the neck are not required unless there are thyroid nodules present.

Cowden syndrome and the PTEN gene



Is Cowden syndrome inherited?
Yes. Cowden syndrome is a hereditary cancer syndrome caused by a germline mutation in the PTEN gene.

There is a 50% chance of a person who carries a germline PTEN mutation, whether male or female, passing the mutation to their son or daughter. If a mutation is identified, then predictive testing is available for blood relatives and is Medicare funded.

Does this sound like you or your family?
Have you been diagnosed with Cowden syndrome or has a PTEN mutation been detected in a blood relative? Genetic testing is available and is Medicare funded.

Make an appointment with Dr Hilda High at Sydney Cancer Genetics. It is a confidential opportunity to discuss your personal and family history of cancer and genetic testing can be organised, if needed.

More information is available on our website, including links to the following:

- PTEN World and PTEN Foundation are support groups for individuals and families affected by PTEN related syndromes. They are based in the USA but have members worldwide.
- The Cancer Genetics section of the Cancer Institute's eviQ website provides up-to-date Australian-based management guidelines
- The US National Library of Medicine website has more information about this syndrome.

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