

## What you should know about Cowden syndrome

Individuals with Cowden syndrome have an increased risk for cancerous and non-cancerous tumors of the thyroid, breast, and endometrium, and an increased risk for colon polyps. Individuals with Cowden syndrome may have some characteristic physical features, including an above average head size (>58 cm in women and > 60 cm in men) and non-cancerous bumps on their skin (trichilemmomas and papillomatous papules).

## The risk for cancer associated with Cowden syndrome

### In women:

- Breast cancer: 25-50%
- Endometrial cancer: 5-10%

### In men and women:

- Thyroid cancer: 3-10% (usually follicular type)

## The risks to family members

Cowden syndrome is caused by mutations in the PTEN gene, and is inherited in an autosomal dominant fashion. This means that children, brothers, sisters, and parents of individuals with Cowden syndrome have a 50% risk to have Cowden syndrome. Individuals with a mutation for Cowden syndrome may develop one cancer, more than one cancer, or none at all.

## Managing the Risk

### General Surveillance:

- Annual physical examination from age 18 with particular attention to the breast and thyroid
- Baseline colonoscopy at age 35 years; repeated every 5-10 years or earlier if indicated

### Breast Cancer Risk:

- Monthly self-examination beginning at age 18 (for females and males)
- Clinical breast examinations every 6-12 months beginning at age 25
- Annual mammography and breast MRI beginning at age 30-35, or five to ten years earlier than the youngest age at diagnosis of breast cancer in the family
- Prophylactic mastectomy to reduce the risk for breast cancer is an option

### Thyroid Cancer Risk:

- Baseline thyroid ultrasound examination at age 18 years
- Annual thyroid examination

### Endometrial Cancer Risk:

- Annual surveillance for endometrial cancer is an option
- Hysterectomy to reduce the risk for endometrial cancer is an option