BC CANCER HEREDITARY CANCER
Provincial Health Services Authority

# *PTEN* Hamartoma Tumour Syndrome (PHTS) Cancer Risks and Management

## Overview

The *PTEN* hamartoma tumor syndrome (PHTS) includes Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome (BRRS), *PTEN*-related Proteus syndrome (PS), and *PTEN*-related Proteus-like syndrome.

PHTS is caused by pathogenic variants in the *PTEN* gene; however, some individuals may receive a clinical diagnosis of PHTS even when a pathogenic *PTEN* variant is not identified.

PHTS is associated with macrocephaly (head circumference ≥ 58 cm in adult females or ≥ 60 cm in adult males), GI hamartomatous polyps, mucocutaneous lesions (either multiple trichilemmomas, acral keratoses, mucocutaneous neuromas, oral papillomas), multiple lipomas, thyroid structural lesions or vascular anomalies, neurodevelopmental conditions (e.g. intellectual disability, autism spectrum disorder), and increased risk for cancer.

This document summarizes the cancer risks and management recommendations for individuals with PHTS.

## Cancer risks associated with PHTS

| Cancer Type | Lifetime Risk |
|---|---|
| Thyroid (especially follicular type) cancer | 10-35% |
| Breast cancer | 40-60% |
| Colorectal cancer | 9% |
| Endometrial cancer | 28% |
| Melanoma | 6% |
| Renal cancer | Up to 30% |

### General

- **Annual complete physical exam** (with focus on neck, breast and skin) and medical history starting at age 18, or 5 years before the youngest *PTEN*-related cancer in the family.

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- Neurological abnormalities on physical exam or evolving neurological abnormalities should prompt consideration of neuroimaging to assess for Lhermitte-Duclos disease.

**Female Breast Cancer**

Breast Cancer Screening:

- Starting at age 18, females should become familiar with the normal look and feel of their breast tissue and to report any changes to their primary care provider promptly. Regular and consistent breast self-exams can support breast self-awareness and are often most effective when done at the end of menstruation.
- **Clinical exam** of the breast and regional nodes every 6-12 months from age 25
- **Annual breast MRI** beginning at age 30 until age 75.
- **Annual mammograms** beginning at age 30 (continue as long as clinically indicated).

Breast Cancer Prevention:

- Discussion of **risk reducing medication** options and review of potential benefits and side effects is recommended. Medications such as tamoxifen, raloxifene, anastrozole and exemestane may reduce the risk of developing a hormone-receptor positive breast cancer.
- **Risk reducing bilateral mastectomy** (RRBM; removing both breasts) reduces the risk of breast cancer by over 90%. The decision to have RRBM is complex and requires discussion regarding benefits and risks of the surgery in the context of a person's general health, life expectancy and personal health beliefs. Routine breast imaging (mammogram and/or breast MRI) is not required after bilateral mastectomy.

**Thyroid Cancer**

- **Annual thyroid palpation** by primary care provider at time of diagnosis, including in childhood.
- Baseline **thyroid ultrasound** at age 5 (or at first diagnosis of PHTS) with annual thyroid ultrasound from age 12.

**Colorectal Cancer**

- **Colonoscopy every 5 years** is recommended starting at age 35, or 5-10 years before the youngest colorectal cancer in the family if that diagnosis occurred before age 40. Frequency may be increased at discretion of colonoscopist based on personal polyp history or symptoms (e.g. rectal bleeding, age at diagnosis, polyp features/hamartomatous polyps).
- There is no data for aspirin use for colorectal cancer prevention in PHTS, but chemoprevention recommendations from Lynch syndrome (a hereditary colorectal

cancer predisposition) can be considered, if not contraindicated: **daily low-dose aspirin** (81 mg; double dose if BMI ≥30) starting 5 years before colonoscopy screening and stopping by age 70 if used solely for colorectal cancer prevention. H. pylori testing and eradication as well as blood pressure control reduce the risk of aspirin-related adverse effects.

**Endometrial Cancer**

- Transvaginal ultrasounds (TVUs), annual endometrial biopsies and pelvic exams are not recommended in British Columbia, as they are either proven ineffective or lack sufficient evidence to support their use for screening purposes.
- Prompt evaluation by a physician is recommended for any unusual uterine bleeding (e.g., bleeding between menstrual periods or any postmenopausal bleeding), or for persistent symptoms such as pelvic or abdominal pain, bloating, increased abdominal girth, early satiety, difficulty eating, or urinary urgency/frequency that are a change from baseline.
- Recommend consultation with a gynecologic oncologist or gynecologist in community to discuss prevention strategies, including **hysterectomy beginning from age 40**.

**Renal Cancer**

- **Renal imaging every six months from age 20**, alternating renal MRI with ultrasound.

**Skin Cancer**

- **Monthly self-examination of the skin** either alone or with the assistance of a relative to look for abnormalities in growth, shape or coloring.
- **Annual clinical skin exam** to support early detection of associated skin lesions. Recommend referral to dermatology for baseline consultation and recommendations, at the discretion of primary care provider.
- **Limit exposure to UV light:** avoid excess sun exposure, wear a hat, sunglasses, and long protective clothing, apply sunscreen with SPF of 30 or more and labeled “broad-spectrum”, and avoid tanning beds and sun lamps.

*Note: In the information above, male/female refers to sex assigned at birth*.

## High Risk Clinic

Individuals with breast tissue who have PHTS or are at 50% risk of having inherited a pathogenic variant in the *PTEN* gene, can be referred to the Hereditary Cancer Program’s

High Risk Screening Clinic for ongoing cancer risk management and decision support. Read more about the High Risk Clinic.

## Family and Reproductive Considerations

### Inheritance

Each child of someone with a *PTEN* pathogenic variant has a 50% chance of inheriting the variant. In BC/Yukon, genetic testing for *PTEN* is available in childhood.

Family members are encouraged to contact their local genetics clinic to learn more about whether genetic testing or cancer screening may be helpful for them. Family members who live in British Columbia or the Yukon can contact our program directly at hereditarycancer@bccancer.bc.ca.

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