BC CANCER COLON SCREENING
Provincial Health Services Authority

# Supporting Patients with Polyposis:
## Information for Health Care Providers

**Polyposis** is defined by numerous polyps detected at colonoscopy and is diagnosed when the cumulative number of polyps removed during a patient's lifetime reaches certain thresholds. Patients with polyposis are at higher risk of developing colon cancer and require more frequent colonoscopy surveillance and may be at higher risk of other types of cancers. Polyposis is categorized by the histology of the polyps: adenoma, serrated lesion or hamartoma.

## What causes polyposis?

For most people, there is no known cause for their polyposis. However, some people are born with a **pathogenic germline variant** (genetic mutation that can be inherited) associated with a **polyposis syndrome**. Patients with polyposis may be eligible for genetic testing to assess whether they have a pathogenic germline variant.

### Adenomatous Polyposis

- 10 or more adenomas
- May be caused by a pathogenic germline variant
- **Screening for extra-colonic cancers required:**
  - Screening for gastric and duodenal (ampullary) cancers
  - Screening for thyroid and skin cancer

#### Examples of Adenomatous Polyposis Syndromes:

APC-Associated Familial Adenomatous Polyposis (FAP)

- Autosomal dominant inheritance
- Two types:
  1. **Classic Familial Adenomatous Polyposis:** > 100 adenomas develop in teenage years with colon cancer by age 30 without colectomy
  2. **Attenuated FAP (AFAP):** < 100 adenomas between ages 40 to 60 years

MUTYH-Associated Adenomatous Polyposis (MAP)

- Autosomal recessive inheritance: Both alleles must be affected (homozygous)
- < 100 adenomas develop between ages 40 to 60 years
- Some patients have serrated polyposis

### Serrated Polyposis Syndrome

- Has either:
  1. 5 or more serrated polyps proximal to the rectum, all ≥ 5 mm, with 2 ≥ 10 mm; **OR**
  2. More than 20 serrated polyps of any size distributed throughout the colon with ≥ 5 proximal to the rectum
- Clinical diagnosis; rarely autosomal dominant inheritance
- May be associated with MUTYH pathogenic germline variant

### Hamartomatous Polyposis Syndromes

- Rare, autosomal dominant inheritance
- Suspect when 2 or more hamartomas identified
- Gene affected, clinical features and risk of extra-colonic cancers vary depending on the syndrome:
  - Peutz-Jeghers Syndrome
  - Juvenile Polyposis Syndrome
  - Cowden's Syndrome
  - Bannayan-Ruvalcaba-Riley Syndrome
  - Hereditary Mixed Polyposis Syndrome

## What do patients with polyposis need to know?

**Polyposis** refers to many polyps found during colonoscopy. There are different types of polyposis depending on the kind of polyp found.

For most people, the cause of polyposis is not known. Some people are born with a change in their genes called **pathogenic germline variants**. These genetic changes can cause polyposis. They are diagnosed with a blood test.

People with polyposis are at higher risk of colon cancer. Regular colonoscopy is required to remove polyps, which can prevent cancer. Screening for other types of cancer may be recommended for people with polyposis.

Close biological relatives of people with polyposis should have a colonoscopy. If there is a pathogenic germline variant found in a family, then biological relatives can also have genetic testing.

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# Recommended Next Steps for Patients with Polyposis

**Patient has colonoscopy and is diagnosed with polyposis**

## Colonoscopist Consultation[1]

Colonoscopist addresses with the patient:

- Their polyposis diagnosis and what it means
- Their colonoscopy surveillance interval
- Whether upper GI cancer screening is recommended
- Whether biological relatives need to be screened
- Whether they are eligible for genetic testing
- Whether they would be eligible for Hereditary Cancer Program (HCP) assessment after genetic testing

## Next Steps Based on Patient's Eligibility for Genetic Testing

Patient is **eligible** for genetic testing

Patient **declines** genetic testing

Patient is **ineligible** for genetic testing

**Colonoscopist orders mainstream genetic testing**
(completes Mainstream Hereditary Cancer Multi-Gene Panel Lab Requisition)

**Colonoscopist provides ongoing care to patient**

**Colonoscopist discusses genetic test result with patient**

Pathogenic variant **found**

Pathogenic variant **not found**

Automatic referral to HCP if ≥ 20 adenomas and/or ≥2 hamartomas

Automatic referral to HCP; Provides recommendations for patient and their biological relatives

## Who is eligible for genetic testing?

Patients who have:

- 10 to 19 adenomas found ≤ 60 years; **OR**
- ≥ 20 adenomas found at any age; **OR**
- Serrated Polyposis Syndrome[2]; **OR**
- ≥ 2 hamartomas

## Who is eligible for HCP evaluation?

Patients who are eligible for and undergo genetic testing are automatically referred to HCP[3] if they have:

- A genetic test result where a pathogenic variant is found[4]; **OR**
- The indication for genetic testing was either:
  - ≥ 20 adenomas found at any age; **or**
  - ≥ 2 hamartomas

[1] The colonoscopist who completed the last colonoscopy for the patient where the patient met the threshold for polyposis will complete the consultation.
[2] Refer to the first page for the Serrated Polyposis Syndrome criteria.
[3] Health care provider does not need to directly refer the patient.
[4] Patients ≤ 60 years with 10 to 19 adenomas, and patients with Serrated Polyposis Syndrome, will be referred for HCP evaluation only if their genetic test result finds a pathogenic variant.

## Helpful Resources

Information about Specialists Ordering Genetic Testing

Lab Requisition: Mainstream Hereditary Cancer Multi-Gene Panel (Genetic Testing)

Video for Patients: Information about Genetic Testing