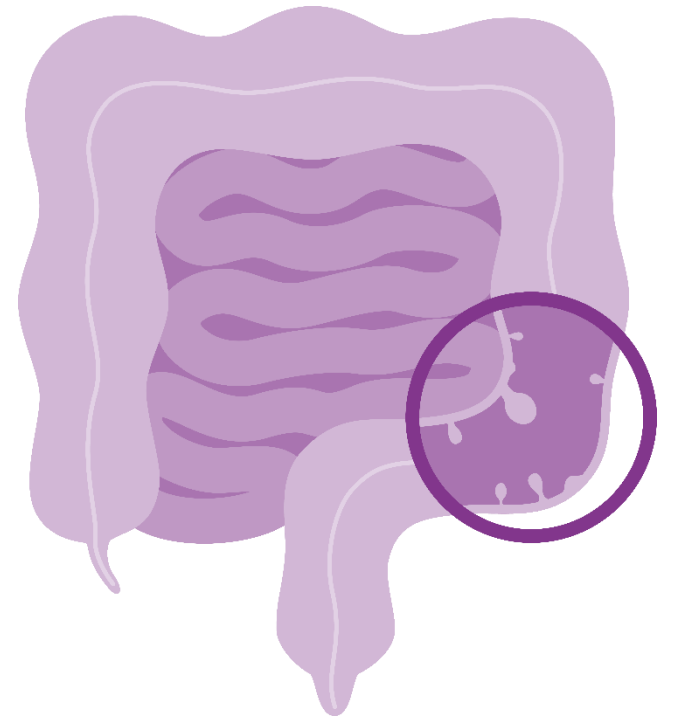
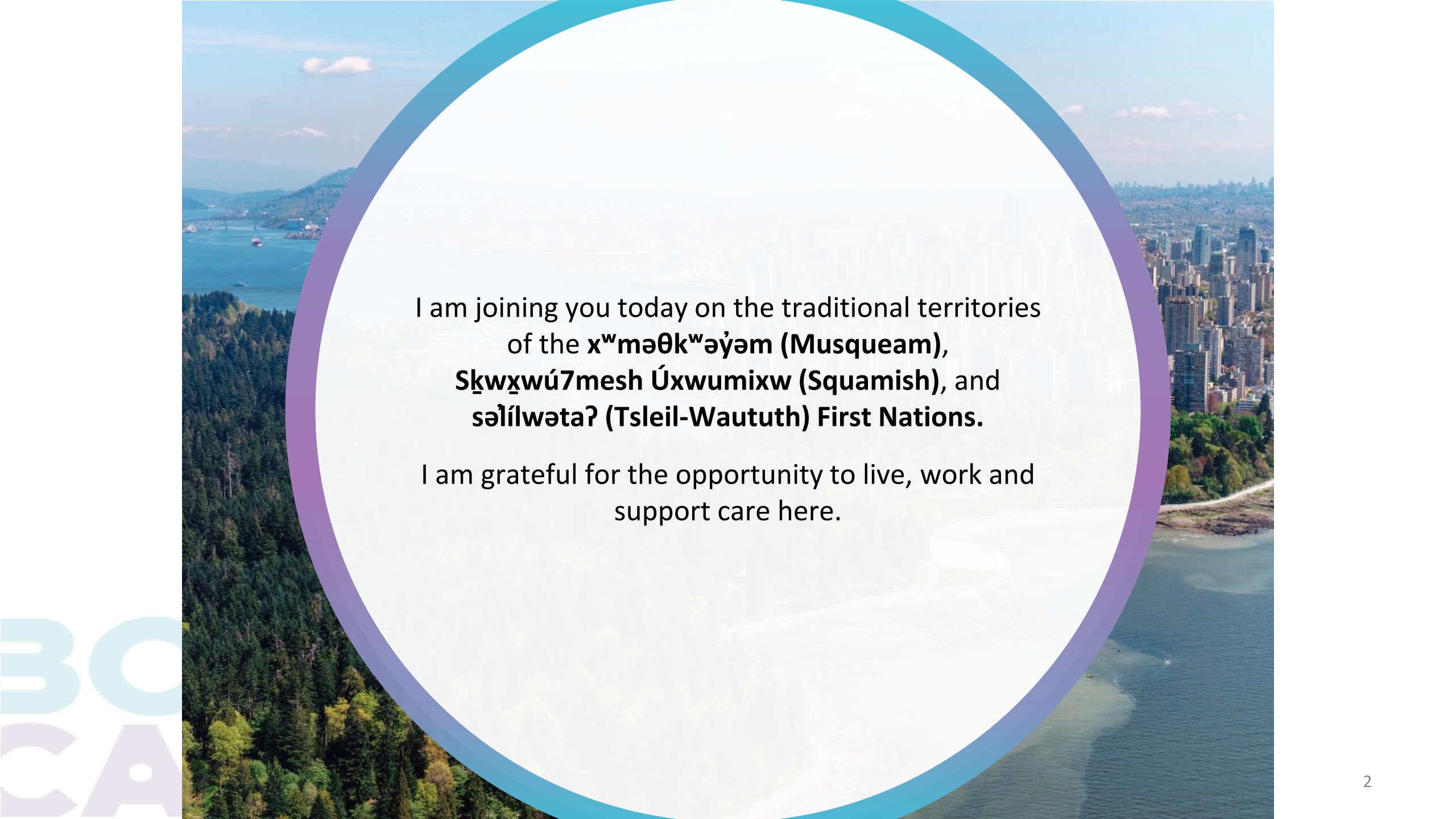


# Polyposis in the Colon Screening Program

Jennifer J. Telford MD MPH FRCPC

Medical Director, BC Colon Screening Program



An aerial photograph of a coastal city, likely Vancouver, showing a dense urban area with high-rise buildings, a large body of water, and forested hills. A large, semi-transparent circular graphic with a blue and purple gradient border is centered over the image. Inside the circle, there is text in English and Indigenous languages.

I am joining you today on the traditional territories  
of the **x<sup>w</sup>məθk<sup>w</sup>əy̓əm (Musqueam)**,  
**Sḵw̓x̓wú7mesh Úxwumixw (Squamish)**, and  
**səlílwətaʔ (Tsleil-Waututh) First Nations**.

I am grateful for the opportunity to live, work and  
support care here.

# Objectives

At the end of this session, participants will:

1. Identify patients with polyposis
2. Order mainstream genetic testing on eligible patients
3. Direct colonoscopy surveillance interval for patients with polyposis
4. Arrange other GI cancer screening in eligible patients
5. Discuss screening of first-degree relatives

# Polyposis

- Refers to individuals with multiple colorectal polyps detected
  - Count is cumulative over a patient's lifetime
- Some will have a pathogenic variant on genetic testing
- Increased risk of CRC
- May be increased risk of other cancers, including GI cancers
- Close family members may be at increased risk of CRC

# Polyposis in CSP

- BC Colon Screening Program collects data on surveillance colonoscopy
  - Identify participants meeting criteria for polyposis
- Patient with  $\geq 10$  precancerous lesions on a single colonoscopy
  - Surveillance colonoscopy in 1 year
  - Referral to the HCP for genetic testing
- Patient with  $\geq 10$  cumulative precancerous lesions on  $> 1$  colonoscopy
  - Referral to the HCP for genetic testing
  - Surveillance colonoscopy life-long; interval never  $\geq 5$  years

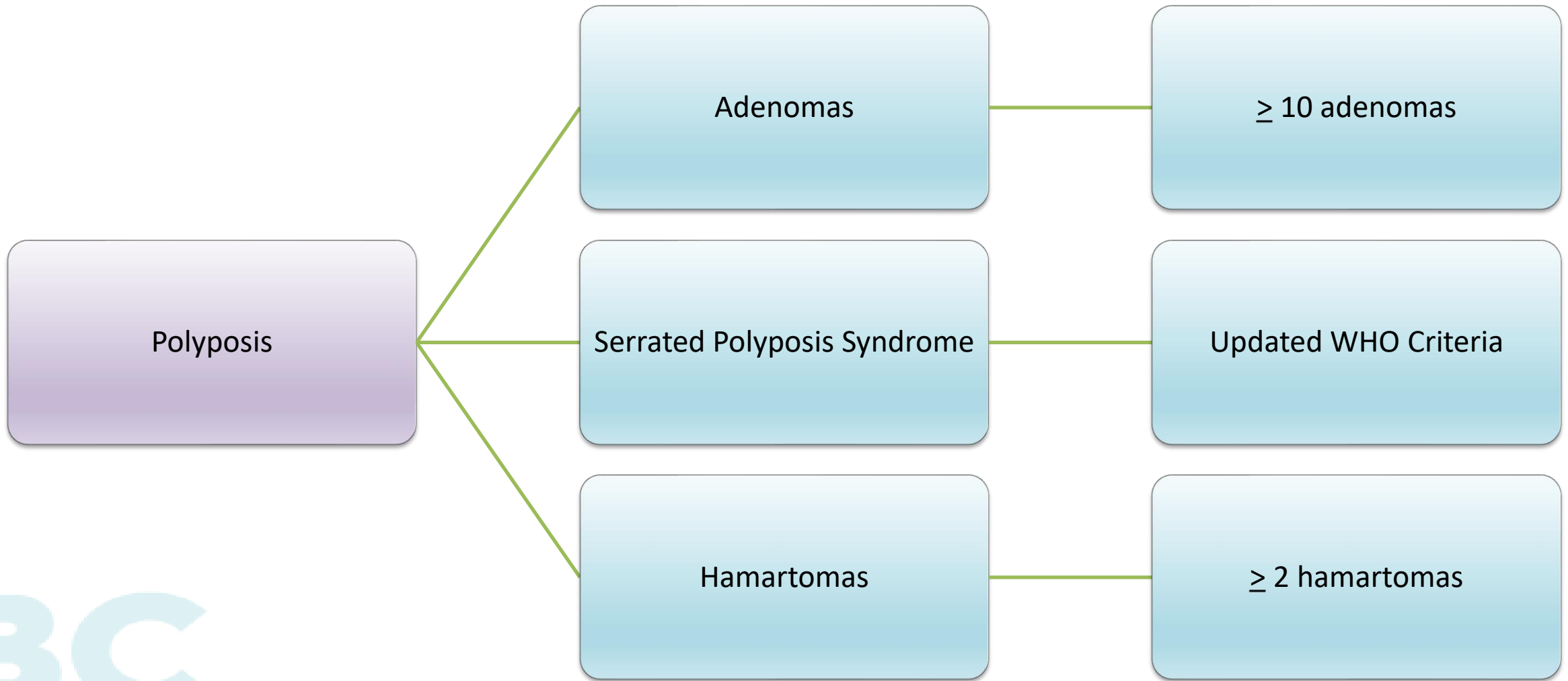
# Polyposis in CSP: Challenges

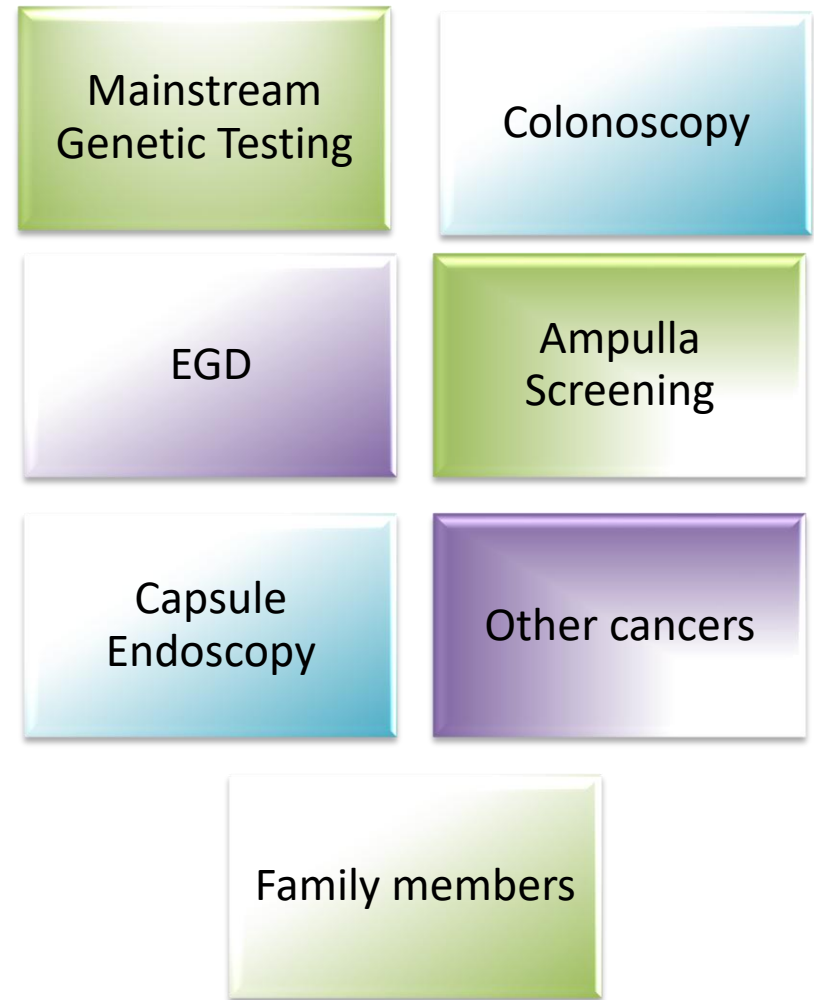
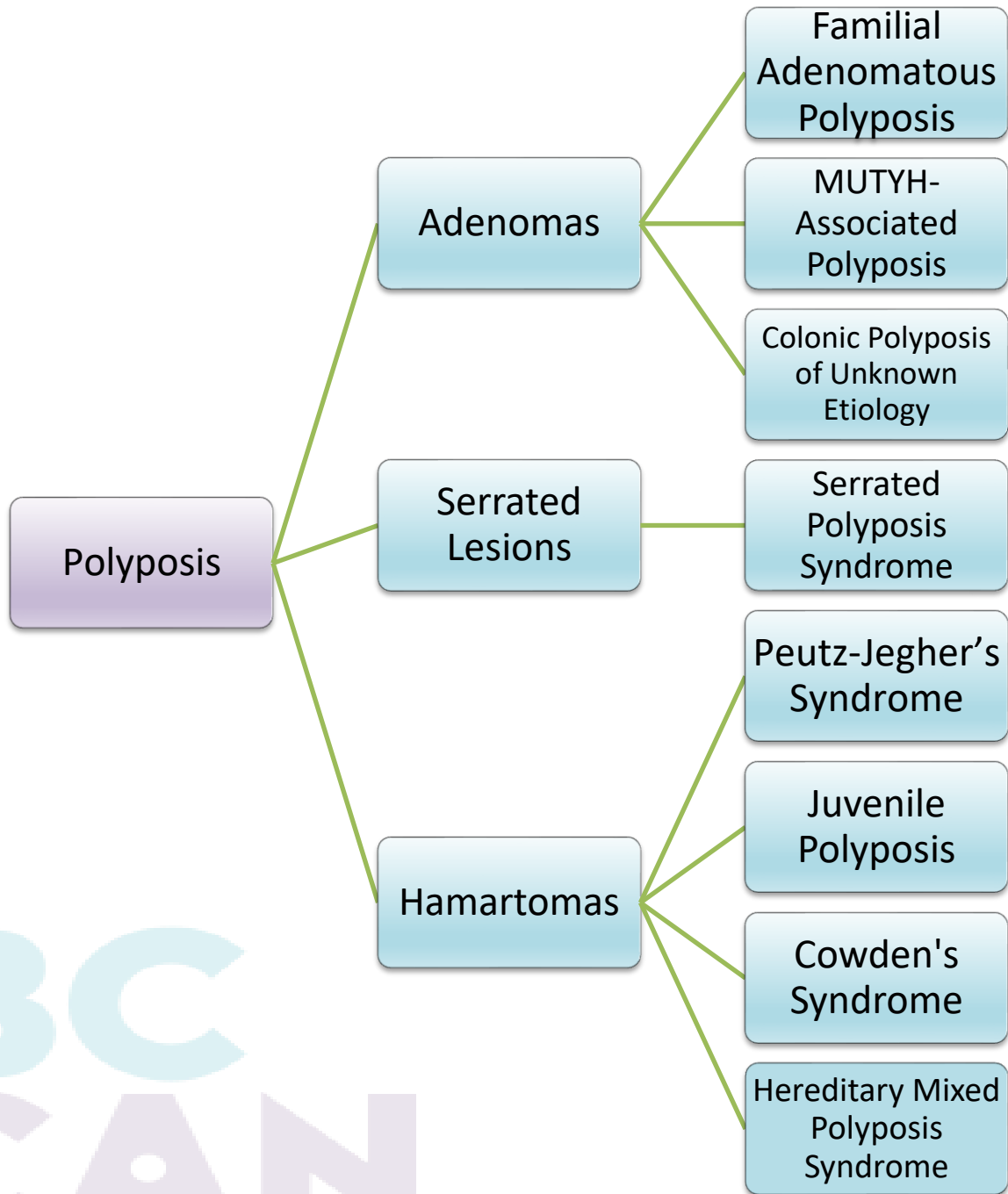
- Missed polyposis diagnoses discovered during the post-colonoscopy CRC review
  - Polyps accumulating over several colonoscopies, different providers
- Program Patient Coordinators may support HCP referral
  - Management strategy for those who decline or are waiting

# Changes to Current State

- CSP is working to provide cumulative polyp counts
  - Total adenoma count
  - Total serrated lesion count with size/location
- HCP has changed eligibility criteria for genetic testing in polyposis
- HCP is enabling mainstream genetic testing for eligible patients

# Polyposis Review





# Familial Adenomatous Polyposis

- Autosomal dominant inheritance
  - ~1/3 do not have a family history
- Germline mutation of APC gene, chromosome 5q21
- Inherit mutation on one allele and acquire mutation or deletion of 2nd allele
- **Classic FAP**
  - Countless adenomas develop in teenage years
  - Develop CRC by age 30 if colectomy not performed
- **Attenuated FAP (AFAP)**
- < 100 adenomas between 40-60 years of age

# Extra-Colonic GI Findings

- Ampullary adenomas and cancer (5% lifetime risk)
- Small bowel adenomas and cancer (4-12% lifetime risk)
- Gastric adenomas and cancer
  - Gastric adenocarcinoma and proximal polyposis (GAPPS)
- Fundic gland polyposis
- Biliary adenomas and cancer
- Abdominal desmoid tumors
- Thyroid cancer
- CNS tumors (medulloblastoma)

# MUTYH-Associated Polyposis (MAP)

- Autosomal recessive inheritance = **need both alleles affected**
- MUTYH encodes a protein that repairs DNA damaged by reactive oxygen species
  - Mutation leads to an accumulation of mismatched base pairs in APC
- Multiple colorectal adenomas (10-100) with high risk of CRC
  - **Also, serrated polyps**
- Age of onset in 40-50s; complete penetrance by 60 years
- Extra-colonic similar to FAP but **possibly** ovarian, bladder, skin and breast

# Colonic Adenomatous Polyposis of Unknown Etiology (CPUE)

- $\geq 10$  adenomas without a PGV identified
- Management depends on number of adenomas
  - $\geq 100$  adenomas = manage as per FAP
  - 20-100 adenomas = manage as per AFAP or MAP
  - **10-19 adenomas = clinical judgement**
    - Begin with colonoscopy Q1-2 years
    - Extend interval out if polyp burden low but **never beyond 5 years**
    - EGD and papilla evaluation Q3 years
    - Extend out interval if normal
- **I use the same strategy for patients who decline testing or are waiting for testing**

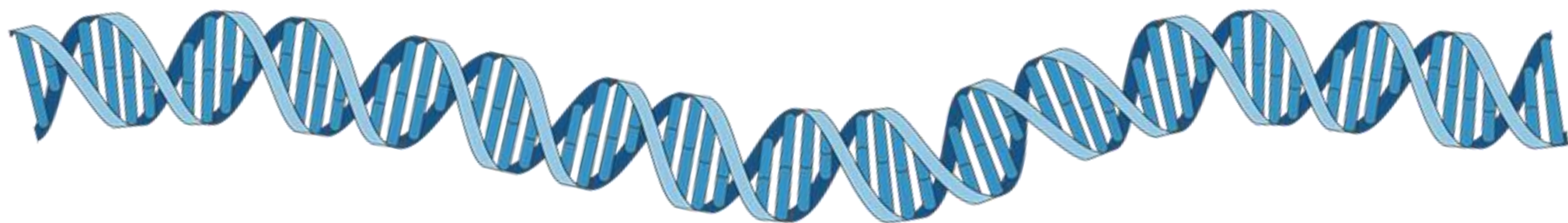
# Serrated Polyposis Syndrome

- WHO Diagnostic Criteria
  1. 5 or more serrated polyps proximal to the rectum, all  $\geq 5$  mm, with 2  $\geq 10$  mm,  
OR
  2. More than 20 serrated polyps of any size distributed throughout the colon with  $\geq 5$  proximal to the rectum
- Often have multiple adenomas as well
- Increased risk of CRC
  - Increased risk associated with a higher number of polyps
  - Wide range in case series (10%-70%)
  - Cancer can occur in small polyps

# Hamartomatous Polyposis Syndromes

Syndrome	Gene	Polyps	Clinical Features
Peutz-Jeghers	STK11	PJ polyp	Cancers: <b>Gastric, small bowel, colorectum, pancreas</b> Mucocutaneous pigmentation
Juvenile Polyposis	SMAD4 BMPR1A	Inflammatory polyp	Cancers: <b>colorectum, gastric</b> Overlap with HHT
Cowden's	PTEN WWP1	Hyperplastic, inflammatory, ganglioneuromas, lipomas, hamartomas, adenomas	Cancers: <b>colorectum</b> , breast, thyroid, gynec, GU Benign skin neoplasms Autism
Hereditary Mixed Polyposis	GREM1	Adenoma, hyperplastic, inflammatory	Cancers: <b>Colorectum</b>

# GENETIC TESTING

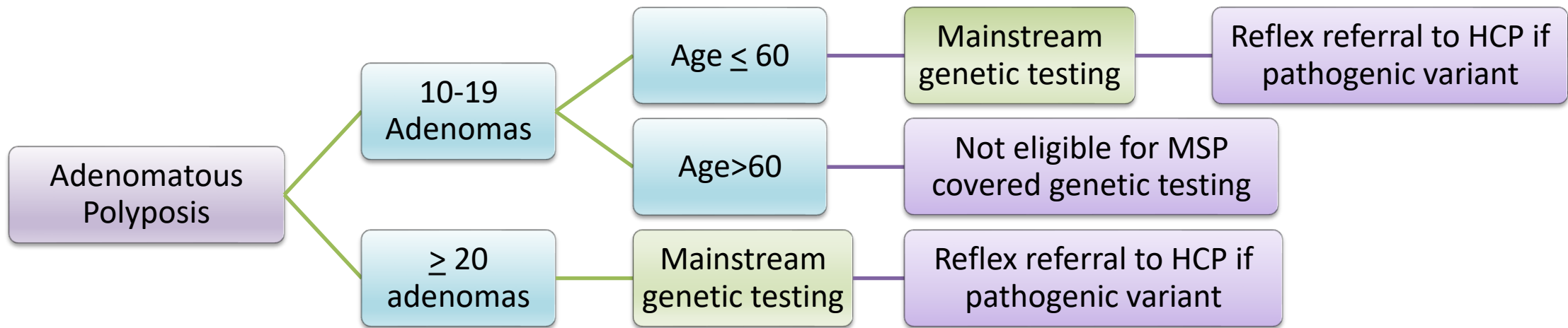


BC  
CAN

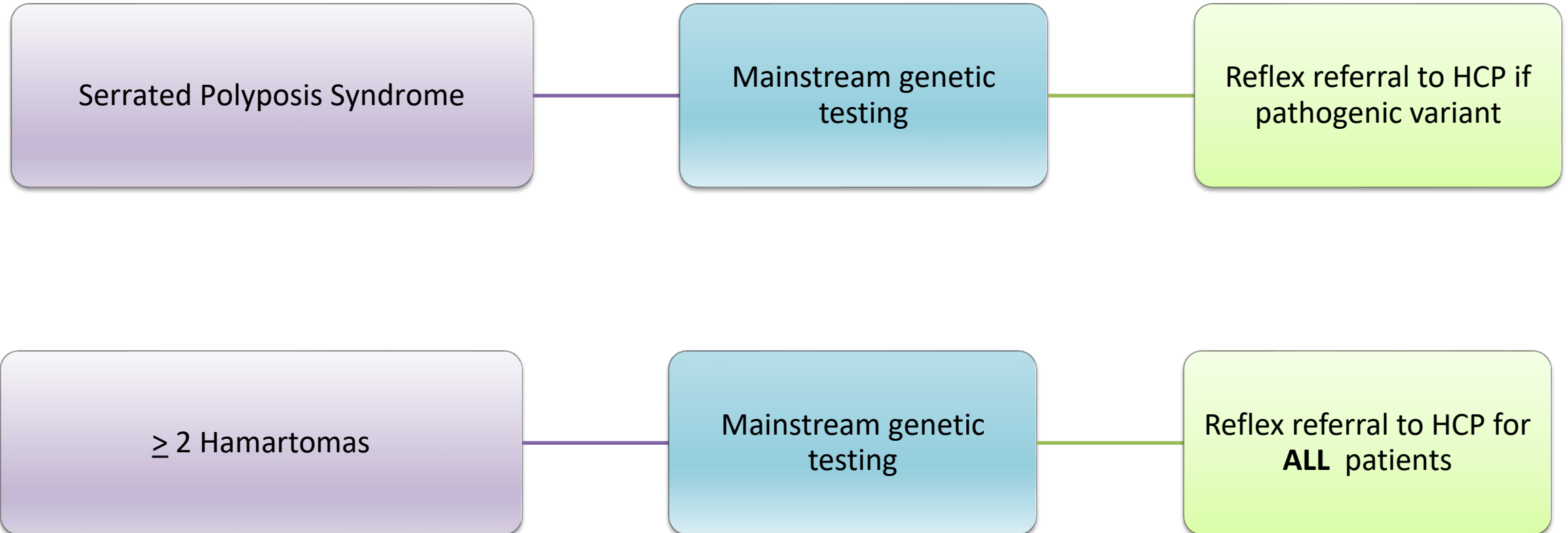
# Mainstream Genetic Testing

- Eligible patient for MSP-covered genetic testing
- Genetic testing ordered by colonoscopist
  - Rec can be downloaded from the Cancer Genetics and Genomics Lab
- Patient can have test done in any BC lab
- Review results with patient
  - Available in 6-8 weeks
- Reflex referral to HCP for positive results

# Eligibility for Testing: Adenomatous Polyposis



# Eligibility for Testing: Serrated & Hamartomatous Polyposis



**CANCER GENETICS AND GENOMICS LABORATORY**

**HEREDITARY CANCER MULTI-GENE PANEL**



BC CANCER  
DEPT. OF PATHOLOGY AND LABORATORY MEDICINE  
ROOM 3307 - 600 WEST 10TH AVENUE  
VANCOUVER BC V5Z-4E6

604-877-6000 ext 67-2094  
FAX: 604-877-6294  
Mon-Fri 8:30AM-4:30PM  
[WWW.CANCERGENETICSLAB.CA](http://WWW.CANCERGENETICSLAB.CA)  
[GENETIC.COUNSELLOR@BCCANCER.BC.CA](mailto:GENETIC.COUNSELLOR@BCCANCER.BC.CA)

CANCER GENETICS LAB  
SHIRE LABEL USE ONLY

PATIENT INFORMATION		REQUESTING PHYSICIAN <small>NOTE: SIGNATURE REQUIRED (below)</small>	
Last Name		Name	
First and Middle Names		MSC	
Date of Birth (dd/mm/yyyy)	Gender <input type="checkbox"/> Male <input type="checkbox"/> Female <input type="checkbox"/> Non Binary/Other/Not Disclosed	Phone	Fax
PHN	BC Cancer ID Cerner MRN	Address	
Email Address		Email Address	
CONSENT		COPY PHYSICIANS (ALL INFORMATION IS NECESSARY)	
Your sample may be sent to a laboratory in the USA for testing. Your personal information (name, date of birth, sex, cancer history) would be sent with the sample. Please contact <a href="mailto:genetic.counselor@bccancer.bc.ca">genetic.counselor@bccancer.bc.ca</a> if you have any questions or concerns.		Name	
Patient agrees to their personal health and genetic information (including test results) being shared with relatives referred for genetic assessment. <input type="checkbox"/> Yes <input type="checkbox"/> No		MSC	
If patient is unable to receive their results, it should be disclosed to (or shared with) Name		Fax - Mandatory to provide copy of genetic test report	
Relationship to patient			
Contact Phone			

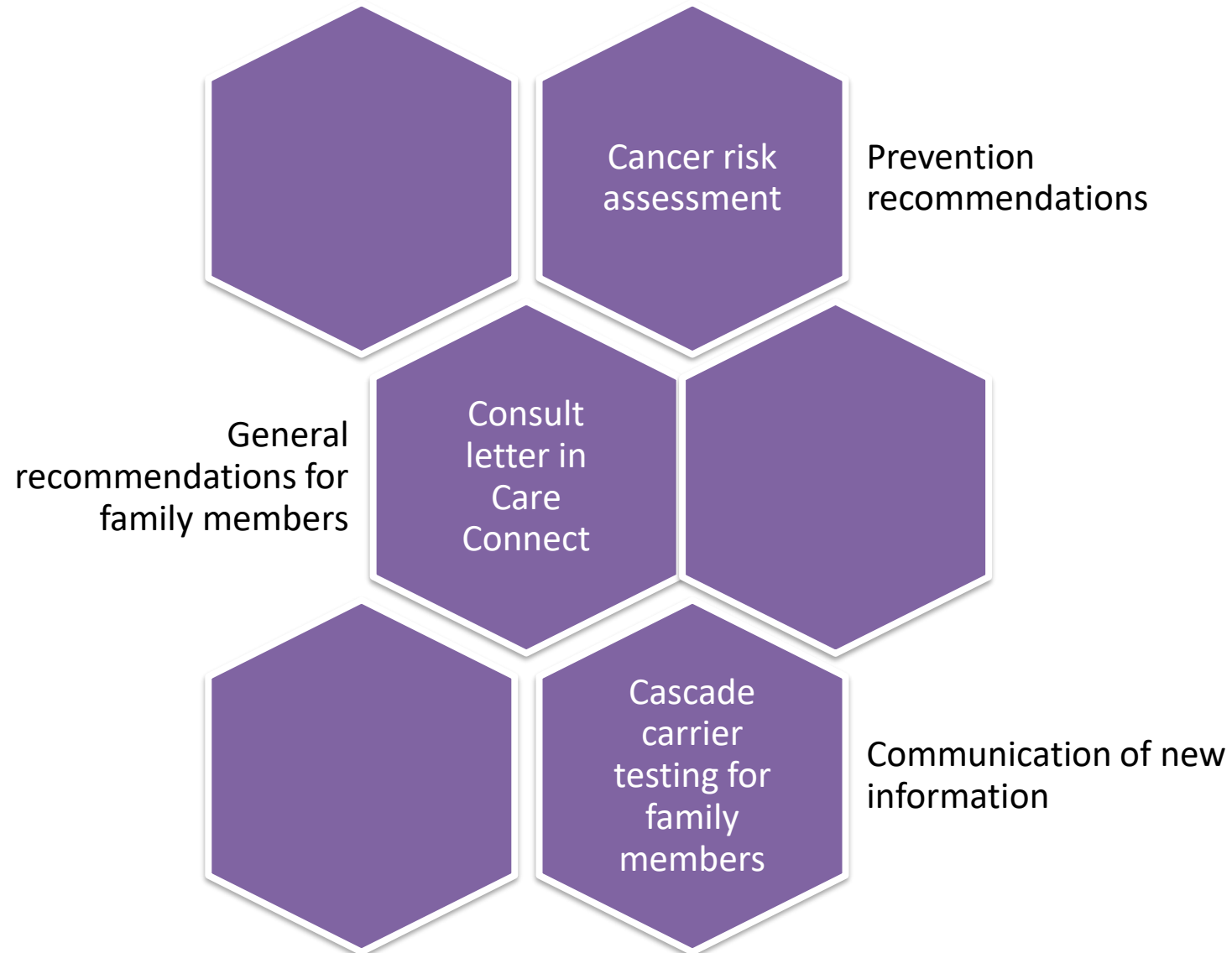
SPECIMEN		HEREDITARY CANCER TESTING INFORMATION	
Specimen Type	Collect 1 x 6mL EDTA blood. <small>Store and ship at room temperature using overnight delivery to Cancer Genetics and Genomics Laboratory (address above). Do not refrigerate or freeze.</small>	<ul style="list-style-type: none"> <li>This is a blood test to see if your cancer is hereditary. About 1 in 10 cancers are hereditary.</li> <li>If your cancer is hereditary, you will have an appointment with a genetic counsellor.</li> <li>Your test results may have implications for relatives.</li> <li>Your test results may be used to guide your cancer treatment and tell us about your cancer.</li> <li>Under the Canadian Genetic Non-Discrimination Act (GNDA), companies (including genetic testing) are prohibited from using your genetic information to discriminate against you.</li> <li>Any unused samples may be stored at the BC Cancer Genetics &amp; Genomics Laboratory.</li> </ul>	
<input checked="" type="checkbox"/> Peripheral Blood	Collection Date	<p><b>TEST</b></p> <p><input checked="" type="checkbox"/> Hereditary Cancer Multi-Gene Panel Testing <small>SO, HCAGPO if your patient</small></p>	
ANCESTRAL BACKGROUND			
Africa / Caribbean	Asia <input type="checkbox"/> East <input type="checkbox"/> South/Central	Europe / UK	Indigenous (First Nations, Metis, Inuit) <input type="checkbox"/> Ash Sep

TESTING INDICATION(S) – SELECT ALL THAT APPLY		
<p><b>Breast Cancer</b> <small>(BRCA)</small></p> <input type="checkbox"/> HER2-negative breast cancer, eligible for adjuvant Olaparib	<p><b>Prostate Cancer</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> Metastatic prostate cancer	<p><b>Ashkenazi Jewish Heritage</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> Personal or family history of breast, ovarian, pancreatic, high-grade prostate cancer
<p><b>Hereditary Breast and Ovarian Cancer</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> Breast cancer ≤ age 35 <input type="checkbox"/> Breast cancer age 36-50 and under active oncologic care <input type="checkbox"/> 2 primary breast cancers at any age <input type="checkbox"/> Triple negative (ER-PR-HER2-) breast cancer <input type="checkbox"/> Ovarian, fallopian tube or peritoneal cancer (non-mucinous epithelial; incl. STIC) <input type="checkbox"/> Male breast cancer	<p><b>Pancreatic Cancer</b> <small>(PANC CA)</small></p> <input type="checkbox"/> Pancreatic ductal adenocarcinoma (PDAC) <i>Does patient have a first degree relative with PDAC?</i> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown <input type="checkbox"/> Pancreatic neuroendocrine tumour	<p><b>Other</b> <small>(INHERCAN)</small></p> <input checked="" type="checkbox"/> ** Approved by Hereditary Cancer Program <input type="checkbox"/> ** Confirmation of pathogenic variant result (include relevant report(s) from tumour testing or clinical trial/research testing) <b>**INDICATION/VARIANT DETAILS (REQUIRED FOR TEST TO PROCEED):</b> 22 adenomas
<p><b>Medullary Thyroid Cancer</b> <small>(MTC)</small></p> <input type="checkbox"/> Medullary thyroid cancer	<p><b>Paraganglioma</b> <small>(PGL)</small></p> <input type="checkbox"/> Paraganglioma (includes pheo)	
<p><b>Renal Cancer</b> <small>(RENAL)</small></p> <input type="checkbox"/> ≤ age 47		

TESTING INDICATION(S) – SELECT ALL THAT APPLY		
<p><b>Breast Cancer</b> <small>(BRCA)</small></p> <input type="checkbox"/> HER2-negative breast cancer, eligible for adjuvant Olaparib	<p><b>Prostate Cancer</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> Metastatic prostate cancer	<p><b>Ashkenazi Jewish Heritage</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> Personal or family history of breast, ovarian, pancreatic, high-grade prostate cancer
<p><b>Hereditary Breast and Ovarian Cancer</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> Breast cancer ≤ age 35 <input type="checkbox"/> Breast cancer age 36-50 and under active oncologic care <input type="checkbox"/> 2 primary breast cancers at any age <input type="checkbox"/> Triple negative (ER-PR-HER2-) breast cancer <input type="checkbox"/> Ovarian, fallopian tube or peritoneal cancer (non-mucinous epithelial; incl. STIC) <input type="checkbox"/> Male breast cancer	<p><b>Pancreatic Cancer</b> <small>(PANC CA)</small></p> <input type="checkbox"/> Pancreatic ductal adenocarcinoma (PDAC) <i>Does patient have a first degree relative with PDAC?</i> <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown <input type="checkbox"/> Pancreatic neuroendocrine tumour	<p><b>Other</b> <small>(INHERCAN)</small></p> <input type="checkbox"/> ** Approved by Hereditary Cancer Program <input type="checkbox"/> ** Confirmation of pathogenic variant result (include relevant report(s) from tumour testing or clinical trial/research testing) <b>**INDICATION/VARIANT DETAILS (REQUIRED FOR TEST TO PROCEED):</b>
<p><b>Medullary Thyroid Cancer</b> <small>(MTC)</small></p> <input type="checkbox"/> Medullary thyroid cancer	<p><b>Paraganglioma</b> <small>(PGL)</small></p> <input type="checkbox"/> Paraganglioma (includes pheo)	
<p><b>Renal Cancer</b> <small>(RENAL)</small></p> <input type="checkbox"/> ≤ age 47		

PHYSICIAN SIGNATURE (REQUIRED)		By signing below, I hereby acknowledge that I have informed the patient about the implications of hereditary testing.	
		DATE	
LAB USE ONLY	PREPARED	HCP USE ONLY	DATE

# Reflex Referral to HCP



# Management of Patients with Polyposis

# Gaps in Management

---

Patients not eligible for genetic testing

---

Patients with a pathogenic variant, awaiting HCP consult

---

Patients who decline genetic testing

---

Patients who do not have a pathogenic variant identified

---

Colonoscopy

EGD

Ampulla Screening

Capsule Endoscopy

Other cancers

Family members

# Risk of CRC and Age to Commence GI Screening

Syndrome	Gene	Lifetime Risk of CRC	Begin Colonoscopy	Colonoscopy Interval (yr)	Begin EGD	EGD Interval (yr)
FAP*	APC	100%	Puberty	1	25-30 yrs	1-3 yr
AFAP	APC	70%	Late teens	1-2	25-30 yrs	1-3 yr
MAP	MUTYH	70-100%	25-30 yrs	1-2	25-30 yrs	1-3 yr
Juvenile Polyposis	SMAD4 BMPR1A	30-40%	Puberty	1-2	Puberty	2-3 yr
Peutz-Jeghers	STK11	40%	8 yrs	1	8 yrs	2-3 yr
Cowden's	PTEN	10-15%	35 yrs	1-2	N/A	N/A
Serrated Polyposis	?	15-35%	40 yrs	1-2	N/A	N/A

\*Flexible sigmoidoscopy until adenomas develop, then colonoscopy

# Colonoscopy Management of Colorectal Polyps

- Surgery for patients with a polyp burden that cannot be managed endoscopically
- Consider if polyp count is  $< 100$
- Frequent colonoscopies (Q3-6 months) until colon cleared
- **Post-operative Management:**
  - After colectomy, survey rectum with polypectomy every 6-12 months
  - After proctocolectomy, survey pouch every 1-3 years

# Management of Gastric Polyps

- Low risk findings = EGD Q 3 years
  - Normal EGD or EGD with fundic gland polyps < 10 mm
- High risk findings = EGD Q 1 year
  - Resect all fundic gland polyps  $\geq$  10 mm
    - Low grade dysplasia does not significantly elevate risk of gastric cancer
  - Resect any antral polyp
    - If concerns for malignant polyp, refer to expert center for ESD
- Consider decreasing surveillance interval
  - Fundic gland polyp with high grade dysplasia
  - Adenocarcinoma resected endoscopically

# Management of Duodenal Polyps: Modified Spigelman's Classification

Factor	Score		
	1 Point	2 Points	3 Points
# Polyps	1-4	5-20	>20
Polyp Size (mm)	1-4	5-10	>10
Histology	Tubular	Tubulovillous	Villous
Dysplasia	Low Grade	--	High Grade

Stage	Points	Surveillance
Stage 0	No polyps	3-5 years
Stage I	1-4	2-3 years
Stage II	5-6	1-3 years
Stage IV	7-8	6-12 months
Stage V	9-12	Surgery

- ❖ Remove all duodenal polyps
- ❖ Capsule endoscopy

# Management of Ampullary Adenoma

- Lesions < 10 mm in size with low-risk histology and appearance can be surveyed
- Endoscopic ampullectomy
  - Lesions > 10 mm in size
  - Advanced histology at biopsy: high grade dysplasia, villous
  - Clinical manifestations
    - Dilated ducts, pancreatitis, elevated liver enzymes
  - No evidence of intra-ductal involvement at EUS
  - No evidence of invasive cancer at EUS
  - Endoscopic appearance consistent with a benign lesion
    - No ulceration
- Surgical resection

# Adenomatous Polyposis without a Pathogenic Variant

- $\geq 100$  adenomas = manage as per FAP
- 20-100 adenomas = manage as per AFAP or MAP
- **10-19 adenomas = clinical judgement**
  - Begin with colonoscopy Q1-2 years
  - Extend interval if polyp burden low but **never beyond 5 years**
  - EGD and papilla evaluation Q3 years
  - Extend interval if normal
  - First degree relatives: colonoscopy at 40 years, Q 5 years

# Serrated Polyposis Syndrome

- Colonoscopy Q 1-2 years
- No extra-colon cancer screening required
- First degree relatives: colonoscopy at 40 years, Q 5 years

# Support for Colonoscopists and Patients



BC  
CAN

# Provider Fact Sheet – Page 1



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

**Polyposis** is when a patient has had 10 or more adenomas, serrated lesions and/or hamartomas in their lifetime. Patients with polyposis are at higher risk of developing colon cancer and require individualized management by a specialist. Some patients with polyposis may be eligible for the Hereditary Cancer Program to evaluate whether they have a genetic predisposition to colon cancer.

### What causes polyposis?

For most people, there is no known cause for their polyposis. However, some people are born with pathogenic variants (changes in their genes) that can cause polyposis syndromes. **Polyposis syndromes** are conditions that cause numerous polyps to grow in the gastrointestinal tract and increase a person's risk for colon cancer and other cancers. Genetic testing can find out if a patient has a polyposis syndrome, such as:

#### APC-Associated Polyposis

- Responsible for 1% of colon cancers
- Autosomal dominant inheritance (about one-third of patients do not have a family history)
- Caused by a pathogenic variant in the APC gene: Inherit mutation on one allele and acquire mutation or deletion of second allele
- Two types:
  - Classic Familial Adenomatous Polyposis (FAP):** Countless adenomas develop in teenage years; Can develop colon cancer by age 30 without colectomy
  - Attenuated FAP (AFAP):** <100 adenomas between ages 40 to 60

#### Serrated Polyposis Syndrome

- Has either:
  - 5 or more serrated polyps proximal to the rectum, all  $\geq 5$  mm, with 2  $\geq 10$  mm; **OR**
  - More than 20 serrated polyps of any size distributed throughout the colon with  $\geq 5$  proximal to the rectum
- Also often has multiple adenomas
- Increased risk of colon cancer associated with a higher number of polyps, including cancer in diminutive polyps

#### MUTYH-Associated Polyposis

- Autosomal recessive inheritance: both alleles need to be affected
- MUTYH encodes a protein involved in DNA repair, and loss of MUTYH results in accumulation of DNA damage
- Causes 10 to 100 colorectal adenomas and/or serrated polyps with high risk of colon cancer
- Starts in ages 40s to 50s, with complete penetrance by age 60
- Extra-colonic features similar to FAP but possibly ovarian, bladder, skin and breast

#### Hamartomatous Polyposis Syndromes

- Peutz-Jeghers:** Caused by pathogenic variant in the STK11 gene; Increases risk of gastric, small bowel, colon and pancreatic cancers
- Juvenile Polyposis:** Caused by pathogenic variant in the SMAD4 and BMPR1A genes; Increases risk of colon and gastric cancers; Overlaps with Hereditary Hemorrhagic Telangiectasia (HHT)
- Cowden:** Caused by pathogenic variant in PTEN genes; Increases risk of colon, breast, thyroid, gynecologic and genitourinary cancers
- Hereditary Mixed Polyposis:** Caused by pathogenic variant in the GREM1 gene; Increases risk of colon cancer

### What do patients with polyposis need to know?

Some people are born with changes in their genes called **pathogenic variants**. Having a pathogenic variant can raise your chances of getting certain types of cancers. It does **not** mean you will get cancer, but it can raise your risk.

Some pathogenic variants can also cause **polyposis**, a condition where many polyps grow in the colon and that can raise your risk of colon cancer and other cancers.

Genetic testing can help find out if you have a change in your genes that is causing polyps to grow in your colon. Genetic testing is done by taking a sample of your blood. The lab uses the sample to check for changes in your genes.

Your genetic testing results can help guide your medical care, including how often you need a colonoscopy. The results can also help your biological relatives choose whether they need to also get tested and manage their cancer risk.

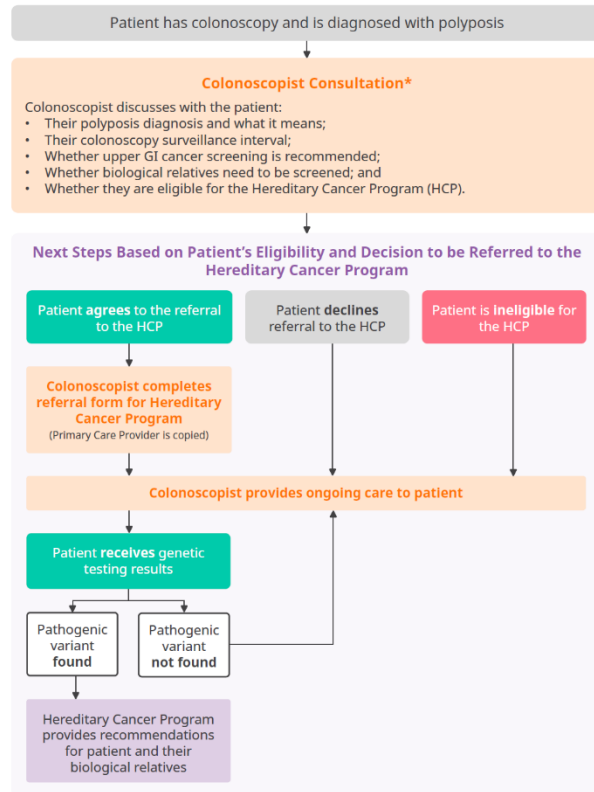
Definition of polyposis

Overview of polyposis syndromes

Key messages for patients

# Provider Fact Sheet – Page 2

## Recommended Next Steps for Patients with Polyposis



**Who is eligible for the Hereditary Cancer Program?**

Patients with polyposis are eligible for the Hereditary Cancer Program if they have had:

- Criteria X
- Criteria X
- Criteria X
- Criteria X

**Who is NOT eligible for the Hereditary Cancer Program?**

Patients with polyposis are not eligible for the Hereditary Cancer Program ...

Next steps for patients with polyposis

Eligibility criteria for mainstream testing

QR code links to mainstream rec and provider information/FAQs

\*The colonoscopist who completed the last colonoscopy for the patient where the patient met the threshold for polyposis will either complete the consultation or refer to another specialist with expertise in polyposis to complete the consultation.

### Helpful Resources



**Health Professionals - Colon Screening**  
[www.bccancer.bc.ca/screening/health-professionals/colon](http://www.bccancer.bc.ca/screening/health-professionals/colon)



**Colonoscopy Standards**  
[www.bccancer.bc.ca/screening/Documents/Colonoscopy-Standards.pdf](http://www.bccancer.bc.ca/screening/Documents/Colonoscopy-Standards.pdf)



**Hereditary Cancer Program Referral Form**  
[https://www.bccancer.bc.ca/coping-and-support-site/Documents/Hereditary%20Cancer%20Program/HCP\\_Form-ReferralForm.pdf](https://www.bccancer.bc.ca/coping-and-support-site/Documents/Hereditary%20Cancer%20Program/HCP_Form-ReferralForm.pdf)

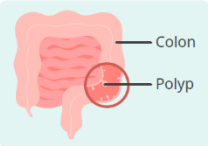
# Patient Fact Sheet: Polyposis and Genetic Testing

**BC CANCER COLON SCREENING**  
Provincial Health Services Authority

## POLYPOSIS AND GENETIC TESTING

**What is polyposis?**  
Polyposis (pronounced *paa-luh-poh-suhs*) is a health condition where many polyps (small bumps) grow in your colon throughout your lifetime.

**What causes polyposis?**  
For most people, we do not know what causes their polyposis. For some people, they are born with changes in their genes called “mutations” or “pathogenic variants”. This means your genes stop working the way they should. Some gene changes can cause **polyposis syndromes**. Polyposis syndromes are conditions where many polyps grow in your colon and that can raise your risk of colon cancer and other types of cancers. Having a gene change does **not** mean you will get cancer — it only makes your chances higher.



**If I have polyposis, what will happen next?**  
Your colonoscopist will talk to you about:

- Your polyposis and what it means.
- How often you should get a colonoscopy and whether you need other types of cancer screening.
- Whether your biological relatives need to also be checked for polyposis and get the follow-up they need. This is because gene changes can be passed down from one generation to another (such as from one of your parents to you).
- Whether you are eligible for genetic testing. **Genetic testing** is done by taking a sample of your blood. The test looks for changes in your genes. It can find out if you have a gene change that may be causing many polyps to grow in your colon.

**Should I get genetic testing? How can it help me and my family?**  
Your colonoscopist will help you decide whether to get genetic testing:

- If you choose to get tested, your colonoscopist will refer you to the Hereditary Cancer Program. The lab report will say whether a “pathogenic variant” was found. The results can help guide your medical care, including what type of cancer screening you should have and how often, tell you about your cancer risk, and whether your biological relatives need to be tested.
- If you choose to **not** get tested, your care will **not** be affected; your colonoscopist will continue to provide you care. You will need regular colonoscopies or other tests to monitor your health. Your colonoscopist will help you decide how often to screen and what type of screening is recommended.

**Where can I get more information?**

- Talk to your colonoscopist or primary care provider.
- Visit the Hereditary Cancer Program’s website: [www.bccancer.bc.ca/our-services/services/hereditary-cancer](http://www.bccancer.bc.ca/our-services/services/hereditary-cancer)

**Questions to ask your colonoscopist:**

- What is polyposis and what does it mean?
- When should I have my next colonoscopy?
- Should I get screening for my upper digestive system (esophagus, stomach and first part of my small intestine)?
- Should I tell my biological relatives about my polyposis so that they can also get checked?
- Am I able to get genetic testing? How can the results help me?
- \_\_\_\_\_

Version: February 2026

What is polyposis?

What causes polyposis?

What will happen next if I have polyposis?

Questions to ask the colonoscopist

How can genetic testing help me and my family? Link to patient video.



# Patient Fact Sheet: Genetic Testing Process

**BC CANCER**  
Provincial Health Services Authority

## GENETIC TESTING: WHAT TO EXPECT

**What is genetic testing?**  
Genetic testing is done by taking a sample of your blood. The test looks for changes in your genes called "mutations" or "pathogenic variants". Changes in your genes can raise your chances of getting some types of cancers, and cancer at a younger age than most people — It does **not** mean you will get cancer.

**How can genetic testing help me and my family?**  
Genetic testing can help you know your cancer risk. The results can also help you and your health care team make choices about your medical care and cancer screening.  
Since gene changes can be passed down from one generation to another (such as from your parent to you), your results can help your biological relatives choose whether they should also get tested and manage their cancer risk.

**My doctor has referred me to the Hereditary Cancer Program. What do I need to do next?**

- 1 The Hereditary Cancer Program will send you a letter with instructions. You choose whether to get more information and get testing through their online portal **or** by phone.
- 2 You register for and go on the secure **online portal**, where you will:
  - Watch videos to learn about genetic testing;
  - Give important information about your health and family history; and
  - Decide whether to have genetic testing (and get started right away if you wish).**Fastest option** **or** If you do not register for the online portal or need an interpreter, the Hereditary Cancer Program will book an appointment by **phone**.
- 3 You get bloodwork done at your local lab.
- 4 Your results will be shared with you by letter or in a phone call with a genetic counsellor. **3 months later**

**Where can I get more information?**  
Visit the Hereditary Cancer Program's website at [www.bccancer.bc.ca/our-services/services/hereditary-cancer](http://www.bccancer.bc.ca/our-services/services/hereditary-cancer).  
If you have questions, contact the Hereditary Cancer Program by emailing [hereditarycancer@bccancer.bc.ca](mailto:hereditarycancer@bccancer.bc.ca) or calling 604-877-6000 (1-800-663-3333) extension 672198.

Version: February 2026

What is genetic testing?

How can genetic testing help me and my family?

What do I need to do after I'm referred to the Hereditary Cancer Program?

# Communicating Information to Providers

BC  
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## BC Medical Journal

CLINICAL

Laura Gentile, RD, MHA, Margot Heintz, BA, RN, CGN(C), Jennifer J. Telford, MD, MPH, FRCPC, CAGF, FACG

### BC Colon Screening Program

Screening for colorectal cancer saves lives. It is more effective when undertaken in an organized screening program.

**ABSTRACT:** Screening for colorectal cancer reduces colorectal cancer-related morbidity, mortality, and incidence. Screening is most effective when administered through an organized program. The BC Colon Screening Program uses a biennial fecal immunochemical test to screen average-risk individuals from 50 to 74 years of age. The program facilitates colonoscopy for those with a positive fecal immunochemical test or as a primary screening strategy for individuals with a high-risk family history. The program is responsible for the technology infrastructure, recalling participants for repeat testing, setting quality standards, and monitoring participant outcomes. A comprehensive quality assurance and improvement program underpins screening activities and includes regular feedback to participating physicians and health authorities.

**Screening for colorectal cancer**  
Colorectal cancer is the third-most-common cancer diagnosis in British Columbia and the second-leading cause of cancer death. It will affect approximately 1 in 14 men and 1 in 16 women during their lifetime.<sup>1</sup>

*Ms Gentile is operations director for the BC Colon Screening Program and the BC Cervix Screening Program. Ms Heintz is a patient coordinator with the Interior Health Authority Colon Screening Program at Penticton Regional Hospital. Dr Telford is a clinical professor of medicine at the University of British Columbia, a gastroenterologist at St. Paul's Hospital, and medical director for the BC Colon Screening Program.*

*This article has been peer reviewed.*

Screening for colorectal cancer detects cancer at an earlier stage of disease, which reduces associated morbidity and mortality and leads to the detection and removal of precancerous colorectal lesions, thereby reducing colorectal cancer incidence. In Canadian modeling studies, several colon screening strategies have been shown to be cost-effective.<sup>2</sup> Screening for colorectal cancer with a biennial fecal occult blood test such as the fecal immunochemical test, preferably conducted through a screening program, is one of the strategies recommended by the Canadian Task Force on Preventive Health Care.<sup>3</sup>

**Why screen for colorectal cancer?**

- It reduces deaths due to colorectal cancer.
- It reduces diagnoses of colorectal cancer.
- It reduces colorectal cancer treatment morbidity (stoma, adjuvant radiation/chemotherapy).
- It is cost-effective.

The best evidence for screening is derived from trials that randomly assign individuals to a control group (no invitation to screen) or to a group that receives an invitation to be screened. **Table 1** presents the pooled results from randomized controlled

trials that assessed annual or biennial guaiac fecal occult blood tests (gFOBTs), 1- or 2-time flexible sigmoidoscopy, and colonoscopy.<sup>4,5</sup> The results are the intention to screen results, which reflect analysis of the entire cohort, whether or not they participated in screening. The period for detecting meaningful differences in colorectal cancer incidence and mortality is at least 10 years. While the meta-analysis of pooled gFOBT trials did not demonstrate a decrease in overall colorectal cancer incidence,<sup>4</sup> there was a reduction in late-stage colorectal cancer incidence: relative risk = 0.92 (95% CI, 0.85-0.99).<sup>3</sup> Colon screening did not reduce all-cause mortality.<sup>3,4</sup>

The gFOBT has been supplanted by the fecal immunochemical test. Several brands are available, which produce either qualitative (positive or negative) or quantitative (mcg globin/g feces) results. Fecal immunochemical tests contain antibodies to human globin, are more specific than gFOBTs, and do not require dietary or medication restrictions. Furthermore, fecal immunochemical tests require a single sample of stool compared with the three specimens required with gFOBTs. These factors have contributed to improved participation in screening

**TABLE 1. Results from randomized controlled trials on colon screening.**

Test	Trial	CRC incidence RR (95% CI)	CRC mortality RR (95% CI)	Follow-up (years)
gFOBT <sup>a</sup>	Pooled results	1.02 (0.93-1.12)	0.91 (0.84-0.98)	19.5
	5 trials	0.90 (0.77-1.04)	0.78 (0.65-0.93)	30.0
Flexible sigmoidoscopy <sup>b</sup>	Pooled results	0.78 (0.74-0.83)	0.74 (0.68-0.80)	11.0-17.0
	4 trials			
Colonoscopy <sup>c</sup>	NordICC trial	0.82 (0.70-0.93)	0.90 (0.64-1.16)	10.0

CRC = colorectal cancer; RR = relative risk; gFOBT = guaiac fecal occult blood test.

## Journal of Family Practice Oncology

By Jennifer J Telford MD MPH FRCPC  
Medical Director, BC Colon Screening Program  
Clinical Professor of Medicine, UBC

Colorectal cancer is the third most diagnosed cancer in BC and the second leading cause of cancer death. However, when detected at an early stage through colon screening, the chances of survival are much higher. Colon screening has proven to be effective in:

- Decreasing incidence of colorectal cancer by detecting and removing precancerous lesions; (Lin et al.)
- Decreasing deaths due to colorectal cancer by detecting cancer earlier at a stage when it is potentially curative; (Lin et al.; Bretthauer et al.) and
- Decreasing morbidity and the need for more invasive surgery, stoma, or adjuvant chemotherapy.

Health care providers play a vital role in supporting patients' participation in colon screening and follow-up care in accordance with the provincial screening guidelines. This includes answering patients' questions about the guidelines and follow-up recommendations. Some of the common questions we have heard from health care providers are:

### What should I tell patients who have had low-risk polyps removed and can now wait 10 years instead of 5 years for their next colonoscopy?

Evidence suggests that people with low-risk precancerous lesions removed from the colon and rectum, are at lower risk of future colorectal cancer than previously thought. (Dubé et al.; He et al.) These individuals can wait 10 years for their next colonoscopy. If no precancerous lesion(s) are found at their next colonoscopy in 10 years, and they do not have a family history of colorectal cancer, it is safe for the patient to return to average risk screening with the fecal immunochemical test (FIT).

Share a copy of the Patient Handout: **Colon Polyps and Follow-Up Recommendations** with your patients who have had precancerous lesion(s) removed during their colonoscopy.



Dr. Jennifer Telford

### What are the colon screening recommendations for patients at higher-than-average risk?

Colonoscopy is the recommended screening test for patients with a high-risk family history of colorectal cancer:

- One first-degree relative (parent, full sibling, child) diagnosed with colorectal cancer diagnosed under the age of 60; or
- Two or more first-degree relatives with colorectal cancer diagnosed at any age

If a patient has a first-degree relative with colorectal cancer, their first screening colonoscopy should be completed at age 40 or 10 years younger than the age of diagnosis of the youngest affected relative (whichever is earliest). For example, if the patient's mother was diagnosed with colon cancer at age 55 and brother was diagnosed with colon cancer at age 40, the patient should have their first colonoscopy at age 30.

Refer to Table 1 for the recommended test, start age, and screening interval for individuals in BC with a family history of colorectal cancer. (Nuk and Telford)

### When can patients stop colon screening?

In BC and across Canada, colon screening

programs stop screening at the age of 75. When the patient turns 75, they will stop receiving reminder letters from the Colon Screening Program. At a population level, people who have been regularly screened with FIT or have undergone colonoscopies may not benefit from screening beyond age 75. Unscreened individuals or those who are not up to date with screening should be offered at least one time screening even if they are over 75 years of age. (Cenin et al.)

Patients aged 75 and older can discuss with a health care provider whether continuing colon screening is the right and safe choice for them. This decision will depend on the patient's personal preferences, their prior screening history, and their overall medical fitness. It is important to recognize that as the patient gets older, the probability of having a complication during a colonoscopy rises.

### Where can I go for more information and point-of-care tools?

Visit [www.bccancer.bc.ca/screening/health-professionals/colon](http://www.bccancer.bc.ca/screening/health-professionals/colon) for colon screening resources specifically developed for health care providers.

To stay informed about the latest resources and updates for health care providers, subscribe to **BC Cancer Screening's Health Care Provider E-Newsletter**. This quarterly e-newsletter highlights resources, tools, and information to help you support patients to prevent cancer and participate in breast, cervix, colon and/or lung cancer screening.

*continued on page 12*

**TABLE 1. BC guidelines for screening individuals with a family history of colorectal cancer.**

Family history	Test	Start age	Interval
≥ 2 FDRs <sup>a</sup> diagnosed with colorectal cancer	Colonoscopy	40 years <sup>b</sup>	5 years
1 FDR diagnosed with colorectal cancer at < 60 years of age	Colonoscopy	40 years <sup>b</sup>	5 years
1 FDR diagnosed with colorectal cancer at ≥ 60 years of age	FIT <sup>c</sup>	50 years	2 years
≥ 1 SDR(s) <sup>d</sup> diagnosed with colorectal cancer	FIT	50 years	2 years
≥ 1 FDR(s) diagnosed with a precancerous lesion	FIT	50 years	2 years

<sup>a</sup> FDR = first-degree relative.

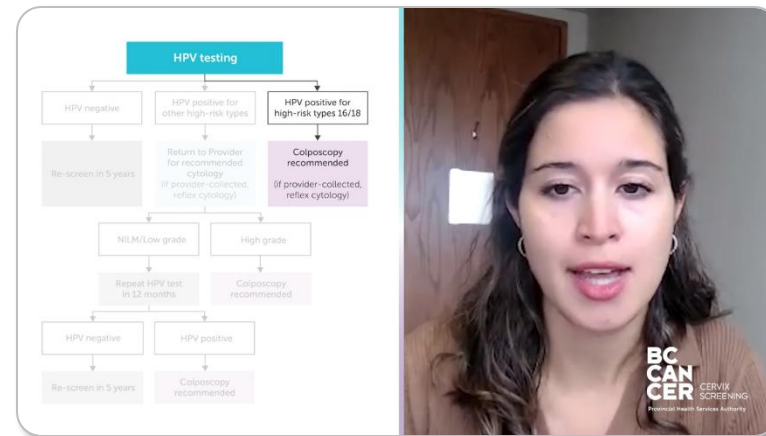
<sup>b</sup> Or 10 years younger than the earliest age of diagnosis of the FDRs, whichever is earlier.

<sup>c</sup> FIT = fecal immunochemical test.

<sup>d</sup> SDR = second-degree relative.

# Video Clips featuring Subject Matter Experts

Drawing on examples from the Cervix Screening Program:



The screenshot shows the Pathways website interface. At the top, there is a navigation bar with the Pathways logo, a home icon, and links for Reports, Resources, Forms, a heart icon, a bell icon, a user profile icon, and a question mark icon. A search bar is located on the right side of the navigation bar. Below the navigation bar, there is a dropdown menu labeled "SELECT SPECIALTY OR SERVICE". The main content area is divided into two columns. The left column contains the main article for the "Colon Screening Program", which includes a title with a heart and envelope icon, a list of related specialties, information about the provider (BC Cancer), a description of the program, eligibility criteria, and a list of individuals not eligible for screening. The right column contains two sections: "Service Types Provided" with "Cancer Screening" listed, and "Ways to Access" with two options: "Provided 1:1 in-person" and "Provided at multiple locations".

**Pathways**™

Home | Reports | Resources | Forms | Heart | Bell | User | ? | Search

SELECT SPECIALTY OR SERVICE

## Colon Screening Program

[Cancer Care](#), [Gastroenterology](#), [Laboratory / Pathology](#), and [Oncology](#).

Provided by **BC Cancer**

A province-wide organized screening initiative designed to detect colon cancer early or prevent it altogether for eligible residents of BC.

Patients with symptoms should be referred directly to a specialist for assessment. FIT is not required. For more information [CLICK HERE](#).

Colon screening guidelines in BC are based on the recommendations by the BC Guidelines and Protocol Advisory Committee (GPAC). For the GPAC recommendations, [CLICK HERE](#).

### SCREENING ELIGIBILITY:

Colon screening (FIT test) is recommended every 2 years to people ages 50-74 with no symptoms. Colon screening (Colonoscopy) is recommended every 5 years for people with at least one of the following:

- One first-degree relative (parent, full sibling, child) with colorectal cancer diagnosed under the age of 60; or,
- Two or more first-degree relatives with colorectal cancer diagnosed at any age.

**Colonoscopy surveillance:** The findings at colonoscopy will determine the timing of further colonoscopies or whether the individual returns to screening with FIT. Patients followed by colonoscopy do not require FIT.

### Individuals NOT eligible for screening:

- Those who are up-to-date for colon screening
- Average risk patients who have had a FIT in the preceding 2 years or colonoscopy or flexible sigmoidoscopy in the preceding 10 years; or
- Higher-than average risk patients who have had a colonoscopy in the preceding 5 years.
- Personal history of colorectal cancer, ulcerative colitis/proctitis or Crohn's disease. Patients should continue to obtain care with their specialist or health care provider
- Individuals who currently have symptoms (e.g. rectal bleeding, persistent change in bowel habits, abdominal pain, unintentional weight loss or iron deficiency anemia). Patients should be referred to a specialist for consideration of colonoscopy, no FIT required



For more information on screening eligibility, please click [HERE](#)

If you cannot find a laboratory in your community, please call us at 1-877-70-COLON (26566).

### Service Types Provided

[Cancer Screening](#)

### Ways to Access

-  Provided 1:1 in-person
-  Provided at multiple locations

# Polyposis Education Session

## Colonoscopy Follow-Up Algorithm

**High Risk Lesions**

- Adenomas with:
  - Villous features
  - High-grade dysplasia
  - $\geq 10\text{mm}$
- Sessile serrated lesions  $\geq 10\text{ mm}$
- Sessile serrated lesions with cytologic dysplasia
- Traditional serrated adenomas
- Hyperplastic polyps  $\geq 10\text{mm}$

Precancerous lesions that do not meet the above criteria are classified as low-risk.

**Low Risk Lesions**

- Tubular adenomas  $<10\text{ mm}$  with low-grade dysplasia
- Sessile serrated lesions  $<10\text{ mm}$  without dysplasia

If the number of precancerous lesions removed during an individual's lifetime is 10 or more, then referral to the Hereditary Cancer Program for evaluation of a potential genetic predisposition to CRC is recommended.

\*Family History and first degree relatives (1st, 2nd or 3rd degree relatives) diagnosis

Telford, Jennifer [BCCancer]

## 14 Days Later: Post-Colonoscopy Assessment

The HA Patient Coordinator:

- Informs Richard of his colonoscopy findings
- Discusses with Richard that he is eligible for assessment with the Hereditary Cancer Program
  - Facilitates the referral after Richard indicates that he would like to proceed
- Establishes that Richard did not have an unplanned medical event while completing the bowel preparation or in the 14 days following colonoscopy

Telford, Jennifer [BCCancer]

[www.screeningbc.ca/health-professionals/colon](http://www.screeningbc.ca/health-professionals/colon)

The screenshot displays the BC Cancer Screening website interface. At the top, the logo "BC Cancer Screening" is on the left, and social media icons for LinkedIn, X, Instagram, and YouTube are on the right, along with a search bar. A navigation menu below the header includes "Breast", "Cervix", "Colon", "Lung", "Health Professionals", and "Contact". The "Health Professionals" menu item is selected, and the breadcrumb "Health Professionals / Colon Screening" is visible. The main heading is "Colon Screening", accompanied by a photo of a doctor and a patient. A sidebar on the right, titled "Health Professionals", contains expandable sections for "Breast Screening", "Cervix Screening", "Colon Screening" (which is expanded to show "Resources" and "Lung Screening"), and "Lung Screening". Below the sidebar, a section titled "Cancer Screening Guidelines" offers "Full Guidelines" with a right-pointing arrow. At the bottom, a "Contact us" link is visible. A teal banner at the bottom of the main content area reads "New to colon screening in BC? What you need to know." and lists three resources: "Program Fact Sheet", "Colon Screening Decision Table", and "Colonoscopy Follow-Up Algorithm".

BC Cancer Screening


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Health Professionals / Colon Screening

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## Colon Screening



### Health Professionals

- Breast Screening +
- Cervix Screening +
- Colon Screening -
  - Resources
  - Lung Screening +

### Cancer Screening Guidelines

Up-to-date guidelines for all four provincial screening programs in one convenient document.

[Full Guidelines >](#)

Contact us

**New to colon screening in BC? What you need to know.**

[Program Fact Sheet](#)  
Colon screening eligibility, guidelines and patient pathway at a glance.

[Colon Screening Decision Table](#)  
A decision-making tool to support health care providers to determine if a patient should have a FIT or colonoscopy.

[Colonoscopy Follow-Up Algorithm](#)

Thank you!

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## FURTHER READING

- Nuk JE and Telford JJ. Familial and Hereditary Colorectal Cancer. BCMJ 2023;65:216-223
- Syngal S et al. ACG recommendations for hereditary GI cancer syndromes. Am J Gastroenterol 2015;110:223-263.
- Dekker E et al. Update on the WHO criteria of serrated polyposis syndrome. Gastroenterology 2020;158:1520-1523.
- Boland CR et al. USMTF recommendations for hamartomatous polyposis syndromes. Am J Gastroenterol 2022;117:846-864.
- NCCN Guidelines