

Hereditary Cancer Update

Mainstream Testing – Who, How, When to Test

Sophie Sun and Kasmintan Schrader

BC Cancer Hereditary Cancer Program Co-Medical Directors
Sept 19 2024

We acknowledge with gratitude, that we are gathered on the traditional, ancestral and unceded territories of the Coast Salish peoples from the x^wm_θθk^w_θy'_θm (Musqueam), Skwxwú7mesh Úxwumixw (Squamish), and səlilw'_θta ¾ (Tsleil-Waututh) First Nations who have nurtured and cared for the lands and waters around us for all time. We give thanks for the opportunity to live, work and support care here

Disclosures

Sophie Sun

Consultant role – AstraZeneca, Bristol-Myers Squib, Roche

Kasmintan Schrader

Consultant role - AstraZeneca, Precision RxDx

Research funding from AstraZeneca and Merck

Medical Advisor relationship and individual stocks and stock options with Genetics Adviser

Co-Inventor on patent application for parent-of-origin determination methodology held by PHSA

Reduce the morbidity and mortality from hereditary cancer syndromes

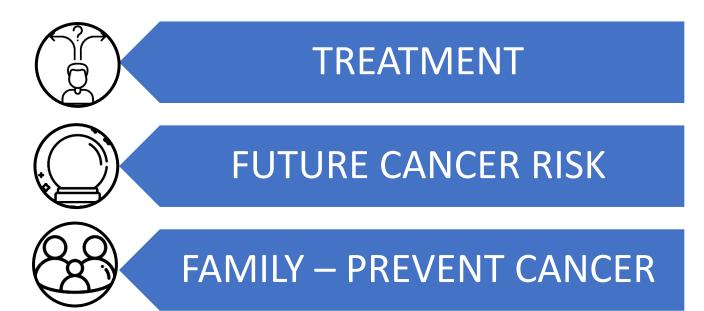
- Identify people with hereditary cancer syndromes
- Cancer risk assessment
- Provide screening and prevention recommendations
- Information to guide cancer treatment decisions
- Identify resources and supports

Genetic Counselling & Genetic Testing

High Risk Clinic

Hereditary Cancer Follow-Up Initiative

Why Test?



How to test?





Cancer Risk Assessment



Screening and Prevention



Information to Guide Treatment



Advocacy and Support



BC Cancer - Screening & Prevention

Staff on site:

- Vancouver
- Abbotsford
- Victoria

Services Provided:

- o Telephone
- Virtual Health
- Small # In-Person

Team:

- Medical Geneticists
- Medical/Surgical Oncologists
- Genetic Counsellors
- Genetic Counselling Assistants
- Nurse Navigators
- Nurse Practitioners
- Clerical Staff
- Research Staff
- Operations Staff





Referrals to HCP



Referral Criteria

- Focus resources on those families more likely to have hereditary cancer
- High demand for services with related waitlist working to reduce this
- Any health care provider or patient can refer to the HCP
- If people don't know their family history, we can often still meet with them
- Family history form not required if patient meets criteria on their own

Increasing Demand

Hereditary Cancer Program (BC & Yukon) Number of Eligible Referrals



Patient Harm

- Missed opportunities to incorporate genetic test results into treatment plans
- Increased rate of patients lost to follow-up
- Increased patient/family anxiety
- Patient death while waiting for an appointment
- 1 Inequitable
- Diagnoses of preventable cancers

Bringing testing into the 'Mainstream'

Oncology Clinic-Based Hereditary Cancer Genetic Testing in a Population-Based Health Care System

- by

 8 Matthew Richardson ¹ □,

 8 Hae Jung Min ² □,

 8 Quan Hong ² □,

 8 Katie Compton ² □,
- Sze Wing Mung ² □, ② Zoe Lohn ² □, ② Jennifer Nuk ² □, ③ Mary McCullum ² □,
- Cheryl Portigal-Todd ² □,
 Aly Karsan ³ □,
 Dean Regier ^{4,5} □,
 Lori A. Brotto ⁶ □,
- Sophie Sun ^{2,7,8,*}

 and

 A. Schrader ^{2,9,10,*}

Cancers 2020, 12(2), 338; https://doi.org/10.3390/cancers12020338



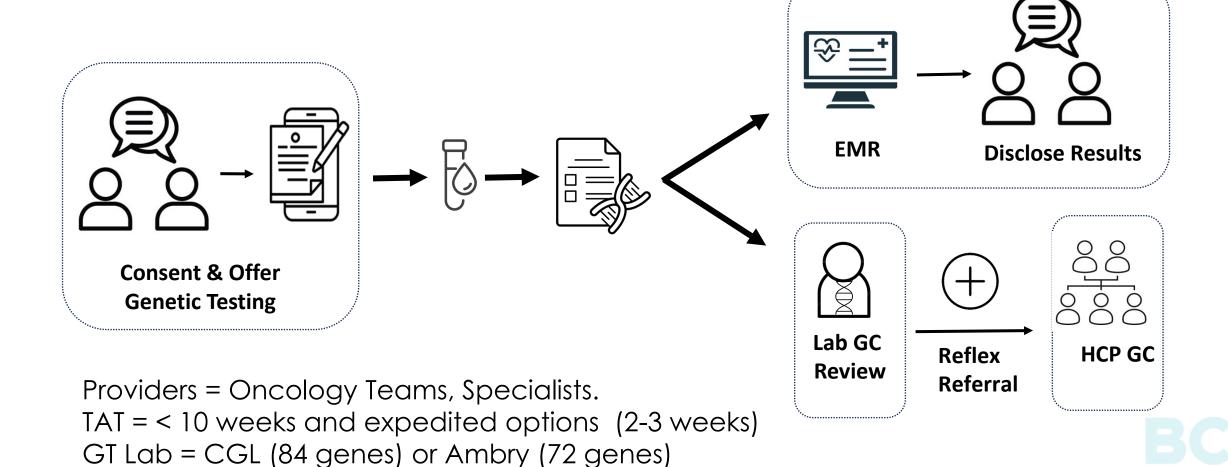
Mainstream 2.0 launched in 2022

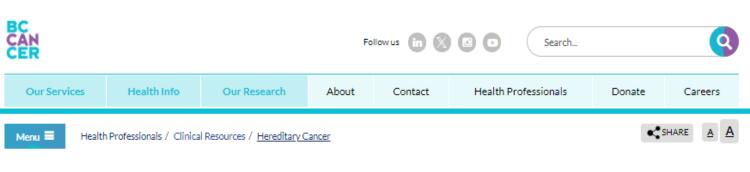






Mainstreaming Process







The Hereditary Cancer Program (HCP) provides genetic counselling and genetic testing for BC/Yukon residents who may have inherited an increased risk for certain types of cancer. Similar services are available across Canada and in other countries.

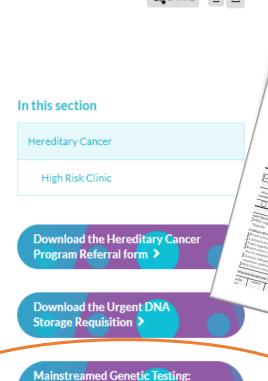
This section provides direction about HCP referrals, information and resources for health professionals to use when discussing hereditary cancer assessment with your patients/families.

Some patients with a personal history of cancer are eligible for "mainstreamed" hereditary cancer testing. You can order the testing and disclose results to your patient without a referral. You can find more information in the "Resources" tab below.

Have a patient in the High Risk Clinic? Learn about your role in their care.

Referral Syndromes Resources

People are encouraged to review their personal and/or family history with a health care provider to clarify whether HCP referral is indicated.



Information and Requisition

Download >

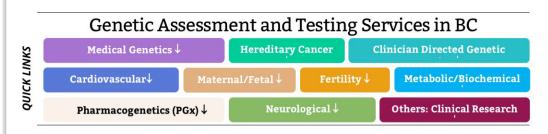




NOW ON PATHWAYS BC

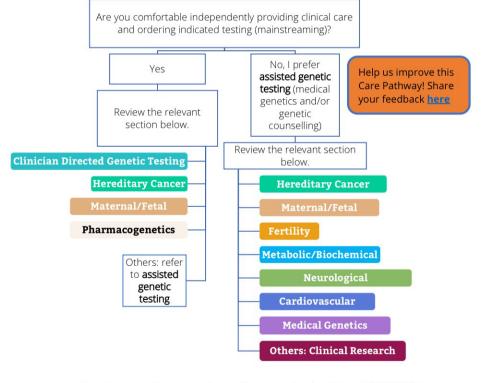
Genetic Clinics listings with estimated wait times & advice options

Expanding Clinician
Tools & Patient Info to
support the use of
genetics in your
practice



This Care Pathway was developed to help healthcare professionals understand when referrals to genetic assessment can change management for patients if there is a suspected genetic basis to the patient presentation/disease. Genetic counselling and assessment and/or testing can be valuable for patients and their families to help them have a better understanding of genetic causes and the inheritance potential of their disease. Pharmacogenetic (PGx) testing can be useful to prevent adverse drug reactions or improve clinical effects in patients with certain genetic variations.

We strongly recommend all care providers to directly order genetic testing (mainstreaming) when possible, to minimize wait times for patients. You will be responsible for ordering tests, reviewing genetic test results, and counselling patients.





Help us improve this care pathway! Share your feedback here <u>survey link</u> Created by Genome BC in partnership with BC genetic clinics. 12 September 2024.

Genetic Assessment and Testing Services Care Pathway

Highlights genetic testing available to be ordered directly by care providers

- Constitutional Genetic tests
- Prenatal Genetic tests
- Hereditary Cancer
- Guidelines & funding available for genetic testing outside of BC

Navigate the referral process Provincial Medical GeneticsSpecialty clinics offering assessment and testing

Help us improve this Care Pathway! Share your feedback **here**

Hereditary Cancer

You can direct order genetic testing (mainstreaming) for **hereditary breast**, **ovarian**, **pancreatic**, **prostate**, **medullary thyroid**, **paraganglioma or renal cancer** for patients whose family history meets the criteria – see clinic page below.

Mainstreamed Hereditary Cancer Testing Clinic

- 1. Review the clinician process:
 - a. Mainstreaming Orientation
 - b. Provider Checklist
- 2. Review genetic testing information with the patient:
 - a. Hereditary Cancer Genetic Testing Patient Information Sheet
 - b. Patient information video
- 3. Fill the requisition form (Genetics and Genomics Lab Hereditary Cancer Multi-Gene Panel) that patients can bring to any laboratory
 - a. http://cancergeneticslab.ca/requisitions/
 - If you use CERNER, enter the "Hereditary Cancer Genetics Panel" order code to note for other providers that you have ordered testing. You will still need to give the requisition to the patient
- 4. (optional) Request advice from a genetic counsellor:
 - a. Contact the <u>BC Cancer Hereditary Cancer Program</u> or request advice using the <u>callback request</u>

Referral to Genetic Testing and Counselling

- 1. Does the patient have: cancer in >1 generation, family member with early onset of cancer (<50), family members having more than 1 type of cancer, or rare cancers?
 - a. Review the <u>referral criteria</u> and consider referral to BC Cancer Hereditary Cancer Program
 - b. If you are still unsure if your patient meets the referral criteria, contact the <u>BC</u>

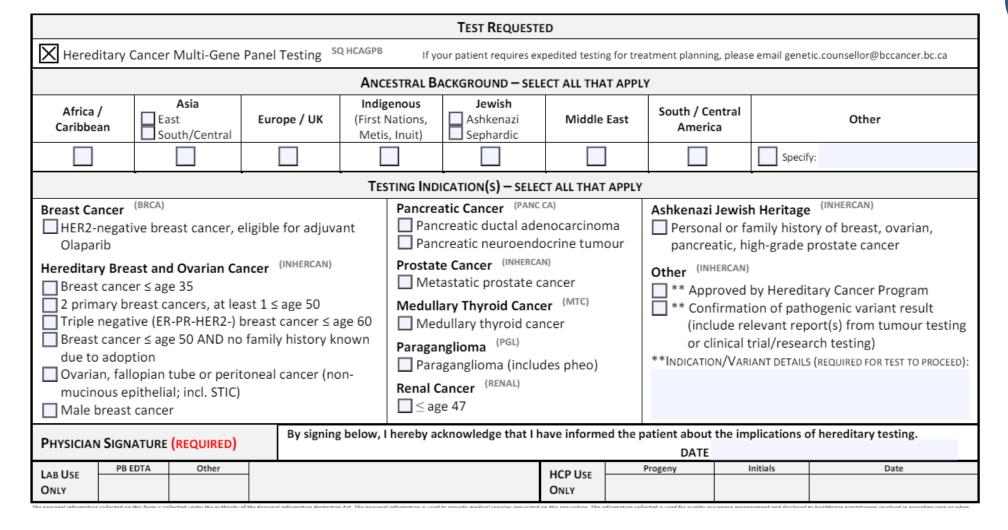
 <u>Cancer Hereditary Cancer Program</u> or request advice using the <u>callback request</u>
- 2. If you have a **pediatric cancer patient**, please refer to **medical genetics**





When and who to test

Who is eligible for mainstream genetic testing?





HCP Mainstream Test Requisition

CANCER GENETICS AND GENOMICS LABORATORY **HEREDITARY CANCER MULTI-GENE PANEL** CANCER GENETICS LAB **BC CANCER** 604-877-6000 EXT 67-2094 SHIRE LABEL USE ONLY DEPT. OF PATHOLOGY AND LABORATORY MEDICINE FAX: 604-877-6294 ROOM 3307 - 600 WEST 10TH AVENUE Mon-Fri 8:30AM-4:30PM VANCOUVER BC V5Z-4E6 WWW.CANCERGENETICSLAB.CA GENETIC.COUNSELLOR@BCCANCER.BC.CA REQUESTING PHYSICIAN NOTE: SIGNATURE REQUIRED (BELOW) PATIENT INFORMATION MSC Last Name First and Middle Names Name Date of Birth (dd/mmm/yyyy) <u>Gen</u>der Phone Fax Male Non Binary/Other/Not Disclosed Female **BC Cancer ID** Address PHN Cerner MRN **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 MSC Name birth, sex, cancer history) would be sent with the sample. Please contact genetic.counsellor@bccancer.bc.ca if you have any questions or concerns. Address Patient agrees to their results being shared with relatives referred to BC Cancer for genetic testing Yes No If patient is unable to receive their results, it should be disclosed to (or shared with): Name MSC Name Relationship to patient Contact Phone / Email Address

HCP Mainstream Test Requisition

CONSENT							
Your sample may be sent to a laborator	ry in the USA for testing. Your p	personal information (name, date of					
birth, sex, cancer history) would be sen	t with the sample.						
Please contact genetic.counsellor@bccancer.bc.ca if you have any questions or concerns.							
Patient agrees to their results being sh Yes No	ared with relatives referred to	BC Cancer for genetic testing					
If patient is unable to receive their results, it should be disclosed to (or shared with):							
Name	Relationship to patient	Contact Phone / Email					

	Specimen		INTERPRETER
Specimen Type	Collect 1 x 6mL EDTA blood.	Collection Date (dd/mmm/yyyy)	Interpreter required?
✓ postalous placet	Store and ship at room temperature using overnight		Yes Language:
Peripheral Blood	delivery to Cancer Genetics and Genomics Laboratory (address above). Do not refrigerate or freeze.		No



Hereditary Cancer Testing

Considerations:



Understanding a factor in my personal / family history of cancer



Learn of other cancer risks, screening and prevention



Information may be used for my current care



Help my family, ie. give options



Increased worry



Strained family relationships



HCP Mainstream Test Requisition

HEREDITARY CANCER TESTING INFORMATION

- This is a blood test to see if your cancer is hereditary. About 1 in 10 cancers are hereditary.
- If your cancer is hereditary, you will have an appointment with a genetic counsellor.
- Your test results may have implications for relatives.
- Your test results may be used to guide your cancer treatment and tell us about new cancer risks.
- Under the Canadian Genetic Non-Discrimination Act (GNDA), companies (including insurers) and employers cannot ask for your genetic test results or ask you to have genetic testing.
- Any unused samples may be stored at the BC Cancer Genetics & Genomics Laboratory and may be used to develop new clinical genetic tests in BC.

TEST REQUESTED									
Hereditary Cancer Multi-Gene Panel Testing SQ HCAGPB If your patient requires expedited testing for treatment planning, please email genetic.counsellor@bccancer.bc.ca									
ANCESTRAL BACKGROUND — SELECT ALL THAT APPLY									
Africa / Caribbean	Asia East South/Central	Europe / UK	Indigenous (First Nations, Metis, Inuit)	Jewish Ashkenazi Sephardic	Middle East	South / Central America	Other		
							Specify:		

Mainstream testing numbers

- $\circ \sim 300 + \text{samples per month}$
- > 1000s Genetic Test Results Reported
- ~12% Positive Rate (~3% recessive/low-penetrant)
- ~25% VUS Rate
 - <2% Result in referral to HCP
- Indication: M-Prostate >> Breast > Pancreatic > Ovarian

Clinician Feedback

'YES, sign me up!'

'Enables access to timely genetic testing for patients (speed of results)'

'Process is straightforward & easy and convenient for patient and ordering providers'

Shifting genetic counselling to after test result for some optimizes genetic services to focus on:

- o Result interpretation, follow-up support for families, cascade testing in relatives
- o Psychotherapeutic genetic counselling to those who need it most
- Mainstream genetic testing increases efficiency and reduces wait times

Genetic test results available to patients, their providers and their families faster.

Resources

BC Cancer Genetics and Genomics Lab Website:

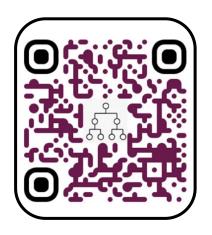
- ☐ Indications
- ☐ Requisition
- □ Process Checklist
- ☐ Patient Information Sheets:
 - Pre-Test
 - Positive Result
 - Negative Result (includes VUS)



Resources

Hereditary Cancer Program Website:

- □Patient video and information sheets
- □Clinician slide deck

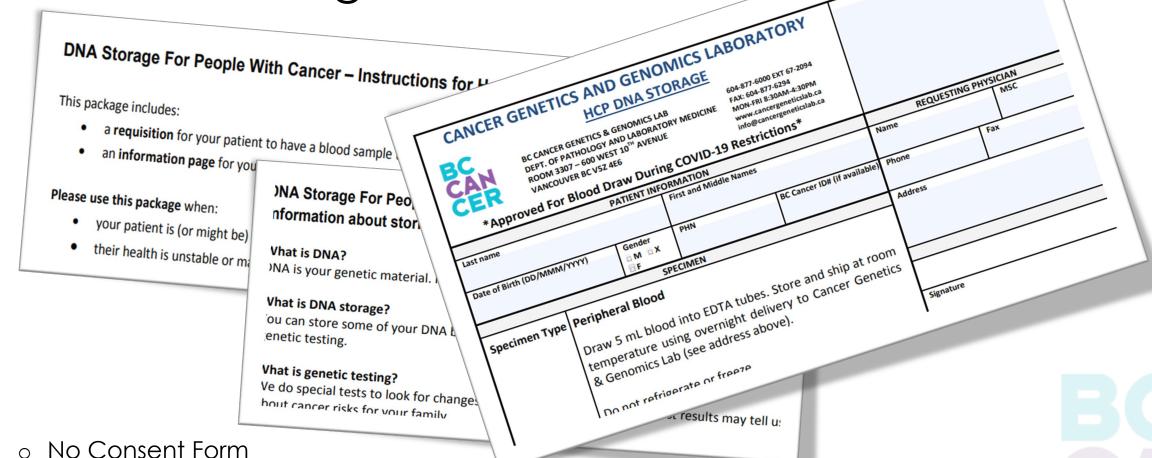




genetic.counsellor@bccancer.bc.ca

DNA Banking

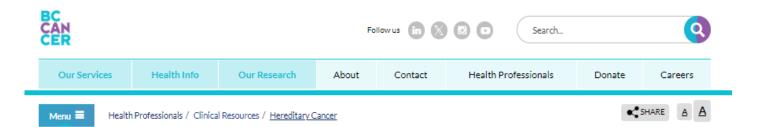
Not an HCP Referral



When to Refer

- Criteria on website
- Patient outside criteria OK to refer
- Any health care provider or patient can refer
- It is OK if people do not know their full family history
- Family history form not required if patient meets criteria on their own
 High demand for services working to reduce waitlist

Who to refer





Hereditary Cancer

The Hereditary Cancer Program (HCP) provides genetic counselling and genetic testing for BC/Yukon residents who may have inherited an increased risk for certain types of cancer. Similar services are available across Canada and in other countries.

This section provides direction about HCP referrals, information and resources for health professionals to use when discussing hereditary cancer assessment with your patients/families.

Some patients with a personal history of cancer are eligible for "mainstreamed" hereditary cancer testing. You can order the testing and disclose results to your patient without a referral. You can find more information in the "Resources" tab below.

Have a patient in the High Risk Clinic? Learn about your role in their care.

Referral Syndromes Resources

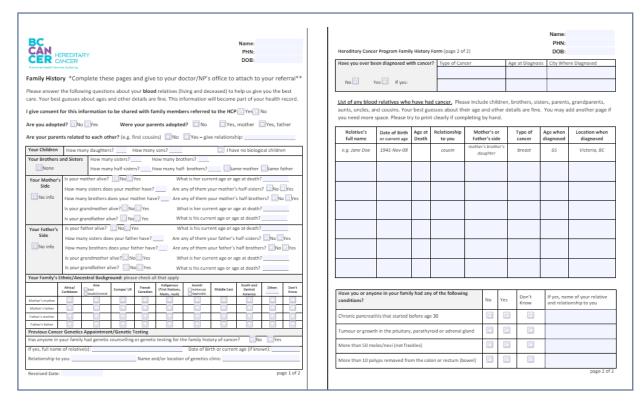
People are encouraged to eview their personal and or family history with a health care provider to clarify whether HCP raterral is indicated.

In this section Hereditary Cancer High Risk Clinic Download the Hereditary Cancer Program Referral form > Download the Urgent DNA Storage Requisition > Mainstreamed Genetic Testing: Information and Requisition Download >



How to Refer

N		Hered	ditary Cancer Program Referral Form							
R	HEREDITARY CANCER	x page 1 (and	completed	Famil	ly History	nages if	required) to:			
Pauli Service Authoriy			ser Health Au		I			kon Health Au	thorition	
ww	w.bccancer.bc.ca/heredita	Y		(F) 604.851.4720		- 1		.707.593		iciionides
DEC	ERRAL DATE:			604.851.4710		74			• 0 local 672198	
		_	1-7							
	erring Clinician :			Billing #:		Phor			Fax:	
Сор	y to/Second Clinician:			Billing #:		Pho			Fanc	
Patient	Personal Health Number			yyyy-mmm-dd)		BC Cancer ID#:		Gender M F X Pronouns:		X
	Last Name	First a	st and Middle Name ty/Town		Phone 1			Phone	2	
_	Address	City/T			Postal Code			Email		
inte	rpreter Required? Ves, la	inguage	:							
Ung	ent Referral? (impact on imp	mediate	cancer	management	or patient i	is pallia	ative):			
	No 🔲 Yes, explain:									
Urg	ent Timeline: <pre>-<1</pre> week	<1 m	onth 🔲	other:				If	patient is ill, s	tore DNA
0	son for Referral – Select 1 o		Cab - C-	Year in direct	·					
								/m		
	ersonal History — attach path e-specific diagnoses:	ology/ot	ner relev	ant report(s) if						
					At least 1 of the following diagnoses at any age: ovarian, fallopian tube or peritoneal cancer (non-mucinous					
	breast cancer s age 35						udes STICI	permunea	Carron (mon-ma	ALTERNA I
	2 primary breast cancers, at les				metastatic prostate cancer					
	triple negative (ER-PR-HER2-)	breast c	ancer sia	ge 60	pancreatic ductal adenocarcinoma					
	breast cancer OR colorectal ca	ncer s as	te 50 ANI	no family	pancreatic neuroendocrine tumour					
	history known due to adoption				Ashkenazi Jewish heritage & personal or family history of breast,					
	colorectal cancer s age 40				ovary, pancreatic, high-grade prostate cancer					
	2 or more colorectal adenoma				male breast cancer					
					=					
	colorectal or endometrial cano				≥ 10 colorectal adenomas (cumulative)					
	2 Lynch syndrome related diag	tnoses, a	t least 1 :	≤ age 50	≥ 2 hamartomatous polyps					
	diffuse gastric cancer ≤ age 50	"additions	I II DGC crit	eria on website	servated polyps meeting WHO 2019 criteria					
	renal cancer ≤ age 47				medullary thyroid cancer					
	billary tract cancer ≤ age 50 *se	Mitineal or	turis on su	Oracida	paraganglioma or pheochromocytoma					
	pathogenic gene variant result – for o					_	_	_		earch testin
	amily History - may include pa									
	a close relative with personal	history:	as selecto	ed above	DEFINITION	NS:				
	breast and ovarian cancer in	dose rek	tives		Breast cance	r: includ	les DCIS an	d excludes t	.CIS	
	2 close female relatives with b				Lynch syndrome related cancers: colorectal, endometrial, ovarian,					
2 close relatives with Lynch syndrome cancer, both s age 50				anth of new 70	gastric, small bowel, hepatobiliary, pancreatic, kidney, ureter, brain					
	3 breast cancers in close female relatives, at least 1 ≤ age 50				tumours, sebaceous gland adenomas, colorectal adenoma 5 age 40					
무	3 or more Lynch syndrome cancers, at least 1 s age 50				Adenomas: tubular or sessile serrated; hyperplastic polyps not included Close relative: children, siblings, parents, aunts, uncles, grandchildren &					
	3 melanomas in close relatives at any age				Close relative: children, siblings, parents, aunts, uncles, grandchildren & grandparents. Can include more distant relatives if appropriate.					
			age		gransparence		Cross III.		auter ii appropria	
			ic variar	nt in family: re	cords requi	ired if	testing d	one outsi	de af BC/Yuko	n
	pproved by Hereditary Cancer I			Relative Nam			ive DOB		How related t	
Ca	arrier Testing - confirmed p									
Gen	e Clinic/City where r	relative t		L	Oakor	diam'	man dan "		h latter days *	
Gen	arrier Testing - confirmed p	relative t			Other In	dicatio	on; descri	be or attac	h letter/medica	l records



Page 1 = Provider

Page 2 & 3 = Patient



Resources

High Risk Clinic

Team:

- Physician
- Nurse Practitioners
- Nurses

Referrals:

- Genetic Counsellor disclosing (+) result
- o GP if Pt discharged from oncology care and has breast tissue
- Pt new to GP practice with variant identified outside of BC



High Risk Clinic

Eligibility

- Not under the care of an oncologist
- People with breasts and a mutation in a gene associated with > 25% lifetime risk for breast cancer (ie, BRCA1/2, ATM, CHEK2, CDH1, PALB2).
- People with Li Fraumeni syndrome (TP53), a syndrome associated with an increased risk of many different cancers.
- People with breasts between ages 30 to 50 with Neurofibromatosis 1 because of increased breast cancer risk.



High Risk Clinic

- Physical Exam
- Screening management for breast cancer: MRI & Mammograms
- Medication for cancer risk reduction
- Prophylactic surgery referral
- Yearly follow-up appointments

Primary care provider for new problems in between visits (like a breast lump or pain or discharge).



Private Pay Testing

Not eligible or do not want to wait for testing Important to choose appropriate lab to ensure accurate results

- Invitae: www.invitae.com/en/cancer
 - requires physician order; \$349USD
- Screen Project: www.womensresearch.ca/active-studies/the-screen-projectstudy/
- Color https://home.color.com/orders/shipping?sku=hereditary%2030(\$260 USD)

If positive, important to refer to HCP for information & ongoing support support (surveillance and prevention; family member testing)









Summary

Hereditary cancer genetic test results can help with:

- Treatment decisions
- Clarifying future cancer risk for a patient and/or their family

Shifting genetic counselling to after test result for some optimizes genetic services to focus on:

- o Result interpretation, follow-up support for families, cascade testing in relatives
- Psychotherapeutic genetic counselling to those who need it most
- Mainstream genetic testing increases efficiency, equitability, and reduces wait times

Genetic test results available to patients, their providers and their families faster.



Who we are

Co-Medical Directors:

Dr. Intan Schrader

Dr. Sophie Sun

Clinical Operations Director:

Adam Kahnamelli

Practice Lead:

Jennifer Nuk

Program Manager:

Mandy Jevon

Aneeta Kassam

Executive Director:

Fabio Feldman

Clinical Coordinator:

Mary-Jill (MJ) Asrat

Clerical Supervisor:

Chelsea Poole

Medical Geneticists:

Dr. Gudrun Aubertin

Dr. Katherine Blood

Dr. My Linh Thibodeau

Genetic Counsellors:

Angela Bedard

Vivian Cheng

Faith Cheung

Genetic Counsellors:

Jacob Coleman

Katie Compton

Courtney Cook

Carol Cremin

Mingshu Dong

Emily Enns

Angela Inglis

Zoe Lohn

Niki Lovick

Allison Mindlin

Tammy Petersen

Ann-Marie Peturson

Genevieve St. Martin

Genetic Counsellors:

Cheryl Portigal-Todd

Manraj Randhawa

Jennifer Thompson

Ruth Turnbull

Genetic Counselling

Assistants:

Alexis Czippel

Kelsey Hamilton

Theo Hui

Suman Kendall

Auto I/h a da ha labah

Aria Khodabakhsh

Lily Teng

Genetic Counselling
Assistants:

Emily Yavorsky

Mir Lafek

Chantel Williams

Clerical Team

Monika Bhatoa

Diane Duyag

Raileen DeLeon

Evangeline Martinez

Jenny Russell

Gagan Sandhu

Ouguii Juliulii

Lhesa Cressey

Krista Oszinski

Data Analyst

Sze Wing Mung

Coop Students

Sasha Abraham

Mitzi Dela Cruz

Clare Heisler

Emma Wong

Cancer Genetics and Genomics Laboratory

Dr. Stephen Yip Dr. Sean Young Dr. Ian Bosdet

Dr. Bahareh Mojarad

Dr. Tracy Tucker

Kristy Dastur



General: HereditaryCancer@bccancer.bc.ca



Mainstream: Genetic.Counsellor@bccancer.bc.ca



High Risk Clinic: HCPHRC@bccancer.bc.ca



1.800.663.3333 local 672198



www.bccancer.bc.ca/hereditary

