HEREDITARY CANCER TESTING IN RELATIVES: PRACTICE GUIDELINES FOR A SUPPORTED DIRECT CONTACT APPROACH TO SHARING INFORMATION IN FAMILIES

Summary of Changes

	NEW	Previous
BC Cancer	New guideline	

1. Introduction

Patients who have had a hereditary cancer risk identified are encouraged to share their result and the availability of genetic testing with their at risk relatives. This provides an opportunity for early detection and prevention of cancer in the family. This Guideline provides an option for certified genetics clinicians and providers to use an evidence-based and patient values-informed approach to increase knowledge about the availability of genetic testing to support families.

This Guideline has been informed by a qualitative research study conducted by the BC Cancer Hereditary Cancer Program exploring the attitudes of people living with hereditary cancer risk in BC/Yukon to a supported direct-contact approach of informing relatives about cascade testing. This approach is designed for actionable conditions where knowledge of a hereditary predisposition will lead to a change in management for family members; however it can also be applied to carrier testing for autosomal recessive conditions or secondary findings that arise from genetic or genomic testing.

1.1. Focus

This document outlines recommended considerations for discussing a supported direct contact approach to facilitating cascade genetic testing with patients and their families.

1.2. Health Organization Site Applicability

This is a Provincial Guideline that applies to all clinical staff providing clinical medical genetics services through the BC Cancer Hereditary Cancer Program.

1.3. Definitions

Cascade Testing: Genetic testing in at-risk relatives after a genetic variant causing disease in a family has been identified.

Cascade Testing allows relatives who test positive to receive tailored surveillance and prevention strategies to decrease their cancer risk. Those who test negative may have reduced anxiety and can be released from increased cancer screening based on familial risk alone.

There are three types of approaches to sharing genetic health information:

1. **Direct Contact:** the genetics clinic contacts relatives directly to tell them about the hereditary condition in the family and their options for genetic testing.

Studies have found this approach can more than double the uptake of cascade testing based on improved communication and understanding of the condition.

2. **Family Mediated:** patient who was initially tested informs their at-risk relatives about their test results and shares a personalized family letter from the genetics clinic to provide information and clinic contact details should that individual wish to have further follow-up.
3. **Supported Direct Contact:** tested patient provides contact information for their relatives and consent for the genetics clinic to communicate with their family members directly.

1.4. Need to Know

The BC Cancer Hereditary Cancer Program is trialing a supported direct contact approach for the next three years. As this is a new guideline and approach to care, clinical cases participating in the supported direct contact approach are continuously compared to a matched direct-contact-naïve clinic population to assess for:

- Time to disease causing variant detection (ie positive genetic test result)
- Uptake of cascade genetic testing
- Overall variant detection rate
- Cost of assessment and testing

2. Practice Guidelines

2.1. Expected Patient/Client/Resident/Family Outcomes

Supported direct contact should be considered as an option to increase information sharing amongst families with hereditary cancer.

Qualitative research has documented positive patient attitudes toward a supported direct contact model. This is seen as one tool to consider and discuss as part of collaborative communication efforts between providers, patients and their families in order to:

- Increase access to cascade genetic testing
- Reduce diagnoses and deaths related to hereditary disease
- Improve the lived experience of the diverse BC/Yukon population accessing medical genetics services

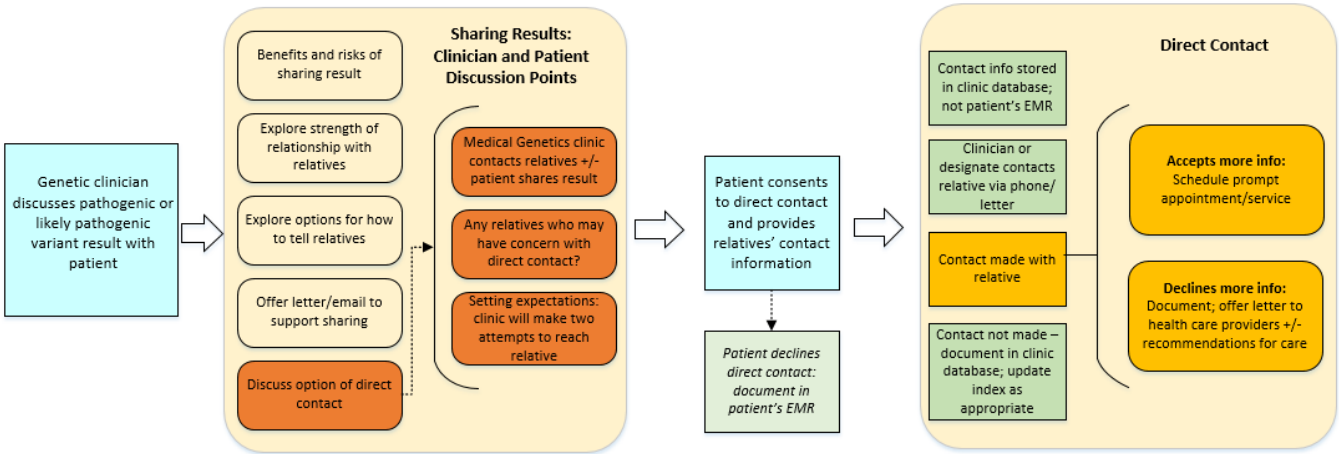
2.2. Assessment

Review of the potential familial implications of genetic testing for hereditary cancer risk should be a part of pre-test information and/or discussions with patients by the ordering clinic or provider.

Review of options for sharing positive genetic test results with at-risk relatives allows for informed shared decision-making with the patient. See section 2.3 for relevant discussion points.

2.3. Intervention

Figure 1: Overview of discussion points and steps in facilitating direct contact with relatives



Information sharing regarding the condition in the family and availability of genetic testing should be generic and focused on the availability of testing to the individual based on a known familial variant. This may help to protect the relative's right "not to know" and to choose whether or not to learn more.

Sample letter template available in section 4: adjust to include the name of the patient who had positive test results depending on patient preference.

Recommend a consultation with PHSA Ethics Service and/or Risk Management for individual cases where there may be complex family dynamics identified or other reasons to invoke a multi-disciplinary approach.

Concerns related to information privacy and confidentiality can be directed to PHSA Privacy Office.

2.4. Documentation

Document the details of the discussion with the patient around supported direct contact of relatives in the patient's electronic medical record.

This information should include:

- Plan for sharing results in family (e.g. patient also intends to share results with family/any specific timeline mentioned, patient will not share results with family/any expressed reasons why)
- Summary of dynamics in family or considerations for timing of contact with relatives
- Date and time of calls or messages left for relatives, dates letter mailed
- Whether or not relative was reached – depending on modality used, if this feedback was requested by patient and with consent of relative if contact successful

2.5. Evaluation

Patient reported outcome measures (PROMs) such as the Genomics Outcome Scale and Multidimensional Impact of Cancer Risk Assessment are in routine use across the clinic. An automated evaluation survey to specifically ask about acceptability and satisfaction around the supported direct contact approach is sent to patients and their relatives.

2.6. Monitoring

Regular review of PROMs and the evaluation survey enables real-time assessment of clinic processes such that modifications can be made if warranted on basis of identification of significant patient distress or dissatisfaction.

Related Document and References

2.7. Related Documents

Template letter to send to relatives – see Appendix, section 4.

2.8. References

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3. Appendices

Appendix 1: Template letter to send to relatives

DATE

Dear Family Member First Name,

Your relative had genetic testing**.** A problem was found in the ___ gene related to a higher chance for ___ cancers. **You are eligible for genetic testing.**

Your family reference number is _____.

What is hereditary cancer?

Hereditary cancer is caused by a gene that is not working. This causes a higher chance for cancer. The gene can be passed down in families.

What is genetic testing?

It is a blood test or a saliva test. It checks for a problem in a gene. Another name for this problem is a gene mutation.

How can genetic testing help you?

If you do not carry the gene change, it is reassuring to know you do not have a higher chance for cancer.
If you do carry the gene change, it is important information for your health. We can help you learn about options to prevent cancer or catch it early.
If you do carry the gene change, it is important information for your family.

Should you get genetic testing?

There are many issues to consider. You have the right to make your own decision about genetic testing. Our program will give you information and support to help you decide. If you meet with us, it does not mean you have to have the test.

We can answer your questions or take a self-referral from you to our program. You do not need a referral from your doctor. We offer services to anyone living in BC or Yukon.

If you live outside of BC or Yukon, we can help you find a program in your area. Similar programs are available across Canada and in other countries.

For more information, contact us or visit our website:

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****Last page of document****

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Posted Date:	21-NOV-2023		
Version:	1.0		
Revision:	**Name of Reviser**	**Description**	**Date**
			DD-MMM-YYYY