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Genetics Education Canada  **Knowledge Organization**

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Mount Sinai Hospital
 Sinai Health System
 Joseph & Wolf Lebovic Health Complex

GEC KO

The Team



Prof. Judith E. Allanson,
MB ChB, FRCP, FRCPC,
FCCMG

GEC-KO co-founder
& co-director
Retired clinical geneticist



Prof. June C. Carroll, MD,
CCFP, FCFP

GEC-KO co-founder
& co-director
Family physician and
clinician scientist



Ms. Shawna Morrison,
MS, CGC

GEC-KO program
manager
Certified genetic
counselor

Challenges & Solutions to Implementation

Challenge

- Securing ongoing funding



Solutions

- Work with experts
- Rely on volunteerism
 - Collaborating with topic experts on educational products and offering authorship
- Form partnerships
 - Research grants that incorporate GEC-KO evaluation or product development

Challenges & Solutions to Implementation

Challenge

- Advocating the value of genetics education to non-genetics health professionals when this is often not viewed as relevant as a stand alone subject

Solutions

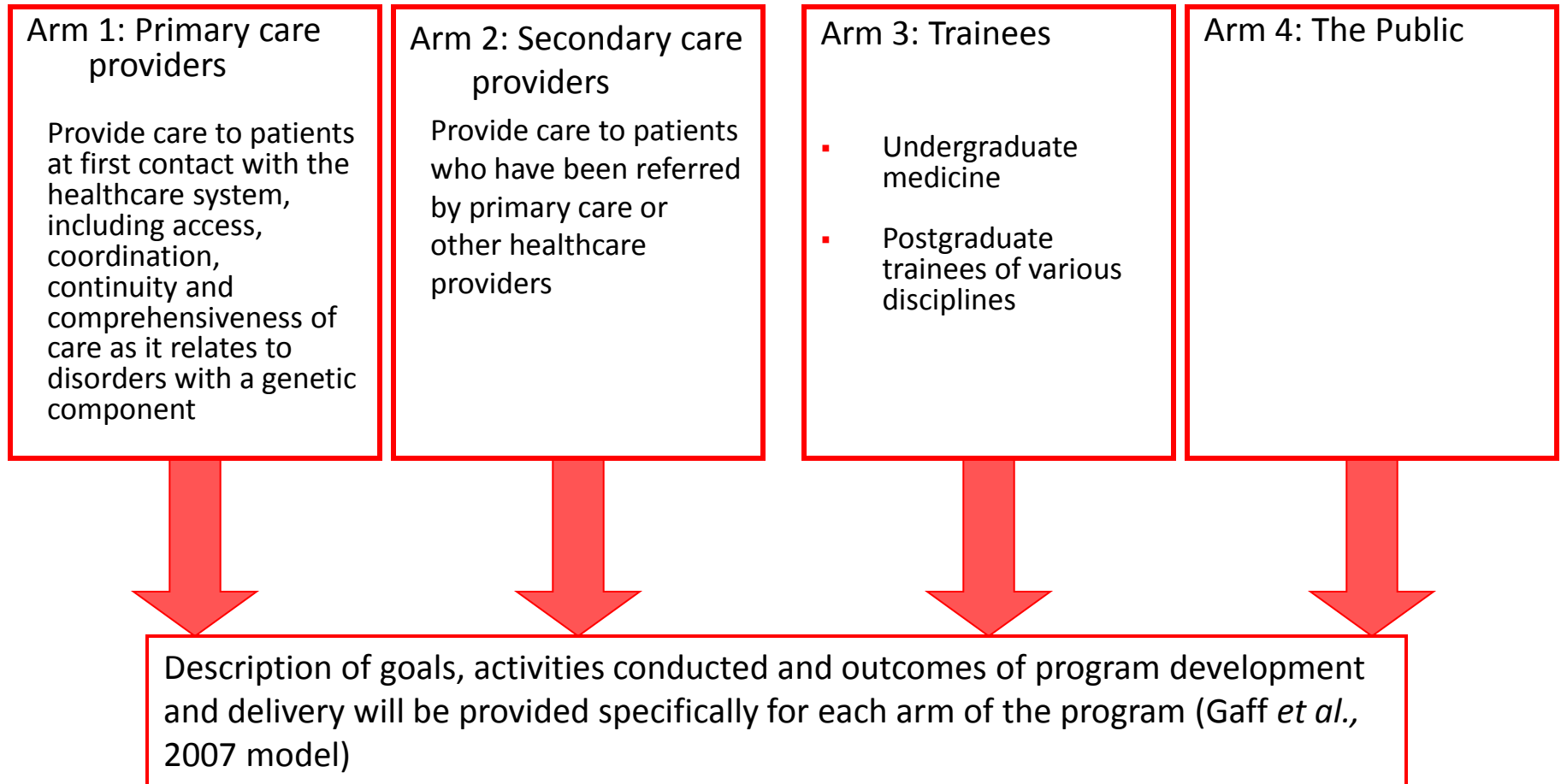
- Integrate in existing well-attended CE venues
- Involve the health professional group in giving the seminar i.e. FP
- Provide ongoing support with resources online and relationships with actual people

Best Practices for Implementation

- Use Program Logic Model
 - Provides clear and purposeful direction, and justification for activities
- Be evidence-based
- Keep resources up-to-date
- Provide resources for point of care
- Evaluate skills wherever possible
- Integrate into existing education venues
- Engage and listen to stakeholders
 - Be flexible, continuously evolve
- Be visible and accessible

Program Map

*All arms relate to provision of genetic services



Goals of Primary Care Arm

Primary care providers will:

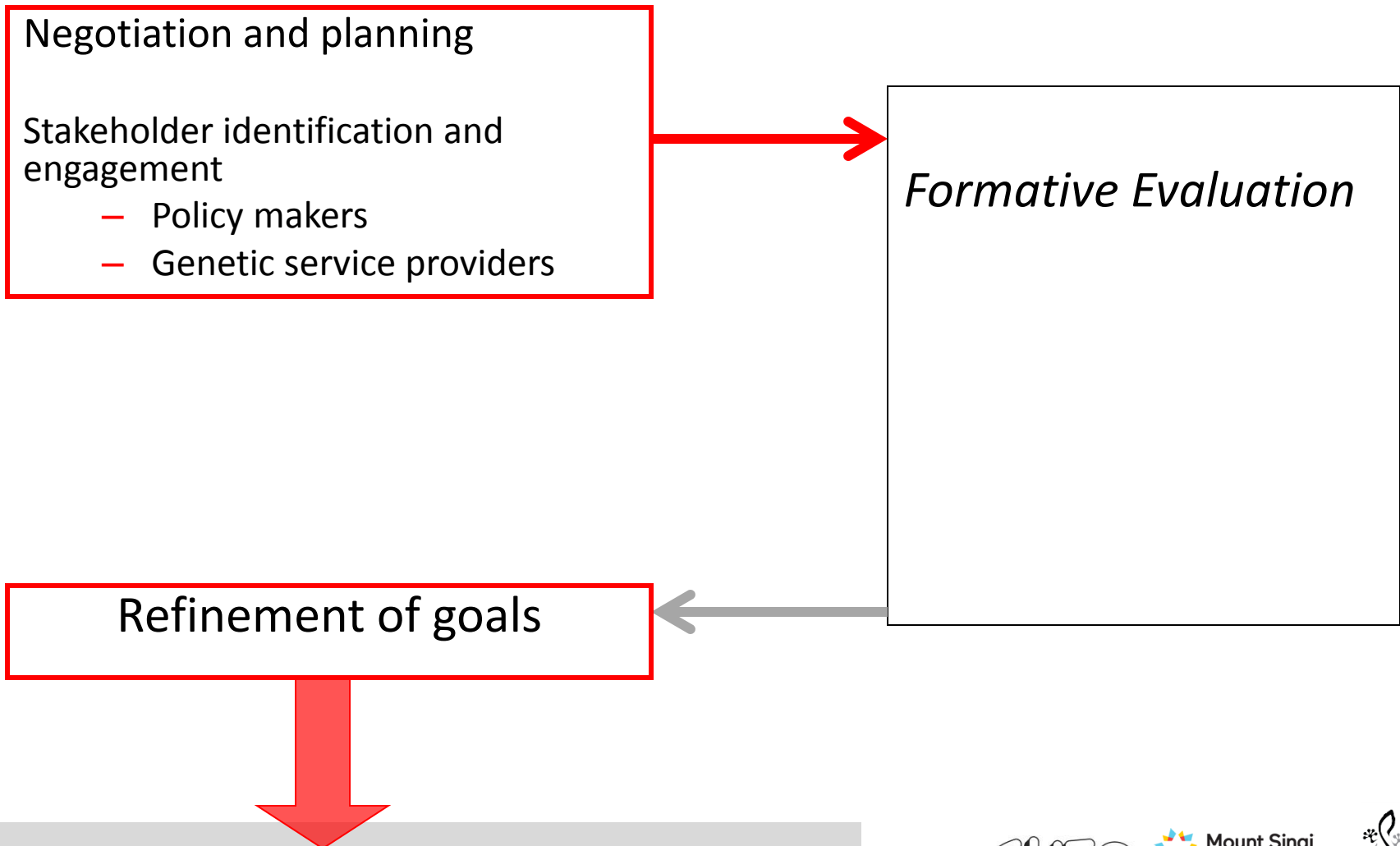
- Have awareness of and use:
 - Genomic educational resources.
 - GEC-KO – website and products.
 - Genomic services and tests.
- Have appropriate genomic knowledge and skills/competencies and confidence in those.
- Demonstrate appropriate behaviour/practice with regards to genomics in primary care.
- Have well-informed attitudes toward the appropriate use of genomic tests and genetics services as related to their practice.

GEC-KO (website and resources) will be considered as having high usefulness, utility, functionality and value.

There will be evidence of improved:

- Quality of care in genomic medicine.
- Management of diseases with a genomic component.
- Improved continuum of care from primary care to secondary care to specialist genetics care.

Awareness of education needs and preferences



Genomic medicine in primary care: Survey Summary

Family physicians have:

An established role in genomic medicine and are optimistic about newer developments

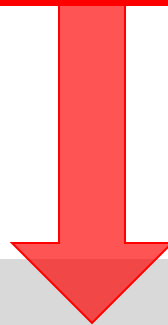
Limited confidence in genomic medicine competencies

High interest in educational resources to enable practice

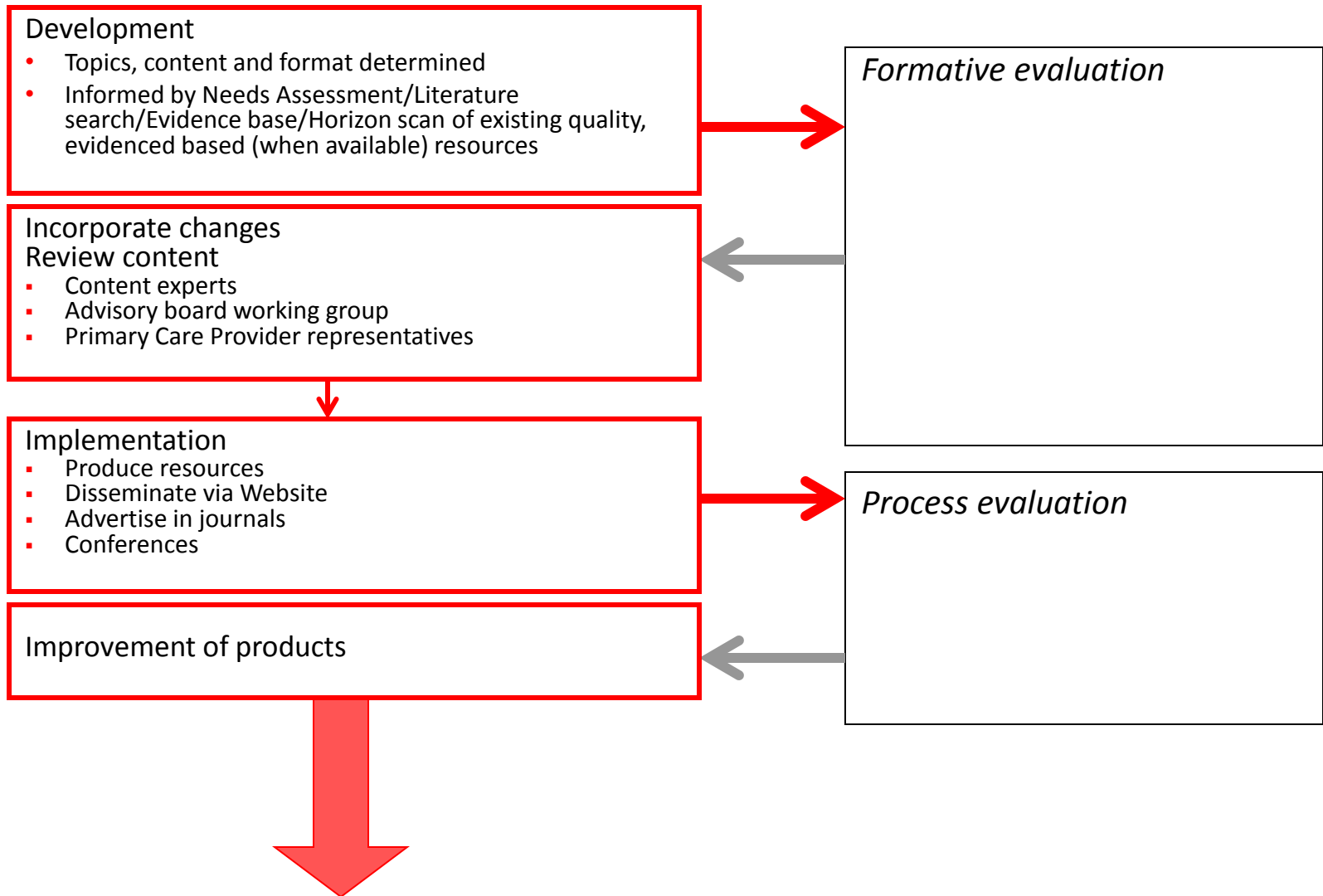
Program Development & Implementation

- Component 1. Written resources
- Component 2. e-Courses/Interactive web teaching & Case-based scenarios
- Component 3. Point of Care (POC) Tools
- Component 4. Website
- Component 5. Models of genetic health service delivery in primary care

**these are tentative components of the program to be modified based on findings from needs assessment*



Component 1: Written Resources





Has the "Angelina Effect" impacted your practice?

Find if your patients are at increased risk for hereditary breast and ovarian cancer caused by changes in the *BRCA1* and *BRCA2* genes with the GEC-KO point of care tool and more.



**BRCA1/2 GEC-KO
on the run**



**BRCA1/2 point of
care tool**



**BRCA1/2 Education
Module**

Canadian Genetics Centre



Find your local genetics centre

Genomics and your practice

Are your patients asking you about private pay genetic testing?

- Yes, all the time
- Fairly often
- Once in a while
- No, never

Family History

Hereditary Cancers

Hereditary Hemochromatosis

Hypertrophic Cardiomyopathy

Ethnicity-based screening in Canada

Factor V Leiden

Use GEC-KO Point of Care Tools to easily and quickly help identify and appropriately refer individuals who may benefit from genetic services at population risk. See the Main Menu for selection.



Part I: Hereditary breast and ovarian cancer referral screening tool to identify patients most likely to benefit from [referral to genetics](#)

Part I of this tool is used to predict which individuals should be referred for genetic counselling due to increased risk for a hereditary breast cancer syndrome including but not limited to hereditary breast and ovarian cancer (HBOC) syndrome caused by mutations in *BRCA1* and *BRCA2* genes. Part II of this tool is used to identify individuals who are at high risk to carry a mutation in *BRCA1* or *BRCA2* genes.

1. Did any of your first degree relatives (parent, sibling, child) have breast or ovarian cancer?	Yes <input type="checkbox"/>	No <input type="checkbox"/>
2. Did any of your relatives have bilateral breast cancer?	Yes <input type="checkbox"/>	No <input type="checkbox"/>
3. Did any man in your family have breast cancer?	Yes <input type="checkbox"/>	No <input type="checkbox"/>
4. Did any woman in your family have breast <i>and</i> ovarian cancer?	Yes <input type="checkbox"/>	No <input type="checkbox"/>
5. Did any woman in your family have breast cancer before the age of 50 years?	Yes <input type="checkbox"/>	No <input type="checkbox"/>
6. Do you have 2 or more relatives with breast <i>and/or</i> ovarian cancer?	Yes <input type="checkbox"/>	No <input type="checkbox"/>
7. Do you have 2 or more relatives with breast <i>and/or</i> bowel cancer?	Yes <input type="checkbox"/>	No <input type="checkbox"/>

Management: **With 1 or more** positive responses, discuss referral to genetics

This POC tool is based on the Family History Screening-7 (FHS-7) (Ashton-Prolla *et al* 2009), which was designed for use in primary care settings and demonstrated an overall sensitivity of 97.0% and a specificity of 53.0% for HBOC syndrome. Overall, **using as cut point one positive answer**, the sensitivity and specificity of the instrument were 87.6% and 56.4%, respectively for hereditary breast cancer syndromes.

Reference: Ashton-Prolla P, Giacomazzi J, Schmidt AV, *et al*. Development and validation of a simple questionnaire for the identification of hereditary breast cancer in primary care. *BMC Cancer* 2009; 9:283
Licence: <http://creativecommons.org/licenses/by/2.0/>



Part II: Red Flags to identify patients at **high risk** of hereditary breast and ovarian cancer most likely to benefit from [referral to genetics](#)

These are general guidelines to identify patients at **high risk** for hereditary breast and ovarian cancer (HBOC) syndrome. You should consider referring your patient to your [local genetics centre or hereditary cancer program](#) for further assessment if s/he has a family or personal history of:

- ▲ Breast cancer diagnosis at a young age (<35-45 years) [both invasive and ductal carcinoma *in situ*]
 - ▲ Ovarian cancer at any age [epithelial]
 - ▲ Male breast cancer
 - ▲ Multiple primaries in the same individual e.g. bilateral breast cancer (particularly if the diagnosis was before age 50), breast and ovarian cancer
 - ▲ Breast cancer diagnosis **AND** a family history of two or more additional HBOC- related cancers, including breast, ovarian, prostate (Gleason ≥7) and pancreatic cancer
 - ▲ High risk ethnicity (Ashkenazi Jewish, Icelandic) and a personal and/or family history of breast, ovarian or pancreatic cancer
 - ▲ Triple negative breast cancer diagnosed <age 60
- OR** if s/he has a personal
- ▲ Probability of 10% or higher to carry a *BRCA* mutation

Eligibility criteria for genetic testing vary among organizations. In general, criteria are based on clinical features that increase the likelihood of a hereditary cancer susceptibility syndrome.

If possible, the affected individual in the family at highest risk to carry a mutation is offered testing first in order to maximize the likelihood of detecting a mutation.

Testing an unaffected individual should only be considered if an affected individual is not available for testing. There are significant limitations to interpretation of test results in an unaffected individual. Unaffected individuals can be referred for genetic counselling, risk assessment and information. It is important to note that any individual of Ashkenazi Jewish ethnicity or French Canadian ethnicities can be offered genetic testing




A library of resources to help integrate relevant genomic information into practice

GEC KO Messenger

Consanguinity

Last updated June 2014

Download the comprehensive  [GEC-KO Messenger](#), the quick reference [GEC-KO on the run](#), and/or the point of care for [ethnicity-based screening in Canada](#). [Link here](#) for an education module with [case-based learning](#).

Bottom line:

Consanguinity is defined as a union between two individuals who are related as second cousins or closer. The chance for adverse outcome in the offspring of a consanguineous union is an estimate based on family history, degree of consanguinity and background population risk. In general, studies have shown that, when there is no known genetic diagnosis in the family, first cousin unions are at a 1.7-2.8% additional risk above the general population risk of 2-3% to have offspring with a congenital anomaly. The risk for a more closely related union is higher and for a more distantly related union is lower. The best tool for counselling a couple about consanguinity is a detailed family history. Genetic testing based on ethnicity, and standard prenatal screening should be offered as for non-related couples. Referral for genetic consultation can be considered if appropriate based on family history and/or screening results.

- > [WHAT IS CONSANGUINITY?](#)
- > [WHO SHOULD BE OFFERED GENETIC TESTING AND/OR REFERRAL?](#)
- > [WHAT DOES CONSANGUINITY MEAN FOR MY PATIENT?](#)
- > [HOW DO I ORDER THE GENETIC TEST?](#)
- > [WHERE DO I REFER MY PATIENT?](#)
- > [RESOURCES FOR HEALTH PROFESSIONALS](#)
- > [RESOURCES FOR PATIENTS AND THE PUBLIC](#)

Authors: S Morrison MS CGC, JC Carroll MD CCFP and JE Allanson MD FRCPC

GEC-KO Messenger is for educational purposes only and should not be used as a substitute for clinical judgement. GEC-KO aims to aid the practicing clinician by providing informed opinions regarding genetic services that have been developed in a rigorous and evidence-based manner. Physicians must use their own clinical judgement in addition to published articles and the information presented herein. GEC-KO assumes no responsibility or liability resulting from the use of information

GEC KO Messenger

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› WHAT IS CONSANGUINITY?

› WHO SHOULD BE OFFERED GENETIC TESTING AND/OR REFERRAL?

› WHAT DOES CONSANGUINITY MEAN FOR MY PATIENT?

✓ HOW DO I ORDER THE GENETIC TEST?

Unless there is a known diagnosis in the family history, likely the only genetic testing offered to your patient will be based on ethnicity.

Ethnicity-based Screening

Certain genetic disorders are more common in populations likely to prefer consanguineous unions (e.g. hemoglobinopathies). Screening for carrier state is recommended in the Canadian Guidelines for Prenatal Diagnosis for individuals belonging to population groups known to have an increased risk for carrying certain genetic disorders. Preconception counselling and testing is recommended in order to arrange for prenatal testing if appropriate.* See the [GEC-KO Point of Care Tool](#) for more on ethnicity-based screening recommendations in Canada.

Hemoglobinopathies

Hemoglobinopathies are a group of inherited disorders that result in abnormal production of the hemoglobin protein due to mutations in the genes responsible for the protein's building blocks, α -globin and/or β -globin.

Thalassemias are due to decreased production of α - or β -globin chains and sickle cell disorders are due to the production of a structurally abnormal β -globin chain. Hemoglobinopathies are common in individuals whose ancestors are from regions where malaria is endemic. **It is recommended that all pregnant women from an ethnic background at increased risk of hemoglobinopathy and/or thalassemia (Table 1) be screened by both CBC, to assess the MCV and MCH, and hemoglobin electrophoresis or high performance liquid chromatography (HPLC).⁵ If both individuals of a couple are**

CONSANGUINITY

Bottom Line: Consanguinity is defined as a union between two individuals who are related as second cousins or closer. The chance for adverse outcome in the offspring of a consanguineous union is an estimate based on family history, degree of consanguinity and background population risk. In general, studies have shown that when there is no known genetic diagnosis in the family, first cousin unions are at a 1.7-2.0x additional risk above the general population risk of 2-3% to have offspring with a congenital anomaly. The risk for a more closely related union is higher and for a more distantly related union is lower. The best test for assessing a couple about consanguinity is a detailed family history. Genetic testing based on ethnicity, and standard prenatal screening should be offered as for non-related couples. Referral for genetic consultation can be considered if appropriate based on family history and/or screening results.

What is Consanguinity?

One billion of the current global population live in communities with a preference for consanguineous union. Consanguinity is defined as a union between two individuals who are related as second cousins or closer.¹

In North Africa, Middle and West Africa, and South Indian populations (and immigrants from these communities) about 20-40% of all unions are consanguineous and first cousin unions account for about 1/3 of all marriages. See Figure 1 for global distribution of consanguinity rates. Reasons for preferring a consanguineous union can include cultural or religious continuity, family solidarity, or reduction of uncertainty associated with health and financial issues. Primary healthcare providers are likely to see couples in consanguineous unions, but these communities who are seeking preconception/prenatal counseling.²

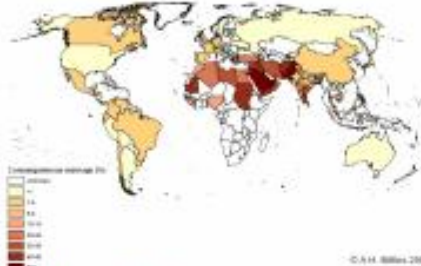
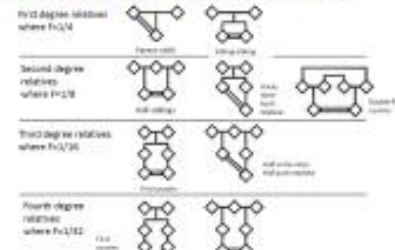


Figure 1. Global distribution of consanguinity rates.²

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WHAT DO I NEED TO KNOW ABOUT THE GENETICS OF CONSANGUINITY?

Inbreeding and consanguinity are terms often used interchangeably, although inbreeding is a regulatory term when applied to humans.³ However, this term is rarely used in population genetics and has some utility. The coefficient of inbreeding (F) provides a numerical risk estimate for the degree of inbreeding of an individual. F values are higher for unions that are more closely related (see Figure 2).⁴ From the thought of an allele proportion of genes in the offspring of a consanguineous union where the allele inherited from each parent is identical (i.e. the proportion of loci at which an individual is homozygous from the same ancestral source).⁵



GEC+KO *on the run*

GEC-KO on the run

Alzheimer disease
Chromosomal microarray
Colorectal Cancer – Lynch syndrome
Consanguinity
Diabetes Type 2
Direct-to-Consumer Genetic Testing
Factor V Leiden – Inherited Thrombophilia
Hereditary Breast and Ovarian Cancer (BRCA1/BRCA2)
Hereditary Hemochromatosis
Huntington Disease
Hypertrophic cardiomyopathy
Non-invasive prenatal testing
Multiple Sclerosis
Pharmacogenomics – Codeine and Breastfeeding
Prenatal Chromosomal Microarray
Schizophrenia

GEC-KO Messengers

Consanguinity
Factor V Leiden – Inherited Thrombophilia

Non-invasive prenatal testing



Download the English PDF [here](#) or download the PDF en francais [here](#). Link here for an education module with case-based learning [here](#).

Non-Invasive Prenatal Testing (NIPT) is a screening test to prenatally detect Down syndrome and other aneuploidies. NIPT assesses fragments of cell-free DNA (cfDNA) that are circulating in maternal blood to determine if there is an increased chance that the fetus has aneuploidy. NIPT should be considered in pregnancies at increased risk of aneuploidy. NIPT has **higher sensitivity and specificity** for Down syndrome (trisomy 21) and trisomy 18 than current screening tests – First Trimester Screening (FTS)/Integrated Prenatal Screening (IPS)/Maternal Serum Screening (MSS) – however it is **not considered to be diagnostic**. Positive results should be confirmed by diagnostic testing (amniocentesis or chorionic villus sampling) prior to any irrevocable action. Negative results may indicate additional follow-up testing and consultation. In Ontario, the Ministry of Health will approve out-of-country funding in certain circumstances. Women who do not meet criteria can pay for NIPT themselves. Price varies by company (~500\$).

Updated Dec 2015

Updated May 2016 *new* Ministry of Healthy Funding for NIPT in [British Columbia](#) and [Ontario](#). Instructions, requisition, links and more below.

WHAT IS NON-INVASIVE PRENATAL TESTING?

Non-invasive prenatal testing (NIPT) is a **highly sensitive and specific** way to **screen** for particular chromosome aneuploidies (an abnormal chromosome number (extra or missing)), in particular trisomies 13, 18 and 21/Down syndrome. NIPT can also be used for sex chromosome identification for the purpose of fetal sex determination where there is increased risk for an X-linked disorder or a sex chromosome abnormality.

NIPT assesses fragments of cell-free DNA (cfDNA) derived from the placenta that are circulating in maternal blood and represent the fetal genetic profile. CfDNA from the pregnancy comprises approximately 10% of DNA in maternal blood and the amount increases with gestational age. Companies offering NIPT use various technologies to analyze cfDNA. Some detect higher relative amounts of DNA from an aneuploid fetus by comparing quantity to a reference chromosome



NON-INVASIVE PRENATAL TESTING

Noninvasive Prenatal Testing (NIPT) is a screening test to potentially detect Down syndrome and other aneuploidies. NIPT assesses fragments of cell-free DNA (cfDNA) that are circulating in maternal blood to determine if there is an increased chance that the fetus has aneuploidy. NIPT should be considered in pregnancies at increased risk of aneuploidy. NIPT has higher sensitivity and specificity for Down syndrome (trisomy 21) and trisomy 18 than current screening tests – First Trimester Screening (FTS)/Integrated Prenatal Screening (IPS)/Maternal Serum Screening (MSS) – however it is not considered to be diagnostic. **Positive** results should be confirmed by diagnostic testing (amniocentesis or chorionic villus sampling) prior to any irreversible action. **Negative** results may indicate additional follow-up testing and consultation. Women who do not meet criteria can pay for NIPT themselves. Price varies by company (750-1-2025).

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NIPT assesses fragments of cell-free DNA (cfDNA) derived from the placenta that are circulating in maternal blood and represent the fetal genetic profile. cfDNA from the pregnancy comprises approximately 10% of DNA in maternal blood and the amount increases with gestational age. Companies offering NIPT use various technologies to analyze cfDNA. Some detect higher relative amounts of DNA from an aneuploid fetus by comparing quantity to a reference chromosome, determining if there is a normal, higher or lower than expected quantity of particular DNA sequences found on select chromosomes (13, 18, 21, X, Y). Others sequence and analyze single-nucleotide polymorphisms (SNPs) to differentiate between maternal and fetal genotypes. NIPT is a noninvasive test performed on a maternal blood sample that poses no risk to pregnancy. Testing can be carried out as early as 9 weeks gestation. A dating ultrasound is recommended prior to drawing the blood sample to ensure viability, obtain an accurate gestational age, and to exclude multiple pregnancies.

NIPT validation studies in high risk populations have demonstrated high pick-up rates/sensitivity for the detection of Down syndrome (sensitivity 99-100 %, trisomy 18 [sensitivity 97-100%], trisomy 13 [sensitivity 79-87%] and sex chromosome differences. False positive rates are reported to be less than 1% overall. Early studies suggest that the positive predictive value (PPV) of NIPT in an unselected, general obstetrical population (low risk) is about 10% for Down syndrome (compared about 0% for standard screening) and about 50% for trisomy 18 (compared about 0% for standard screening). The PPV appears to be significantly higher in high risk populations. A number of women (N=6) have required a repeat blood draw due to initial test failure. Most studies have commercial affiliations.

At the present time, it is recommended that all women under age 35 at estimated date of birth (EDB) be offered prenatal screening, using FTS, IPS or MSS. If a woman is screen positive, NIPT may be considered as a secondary screen of higher sensitivity. Women 35 years or older at EDB can be offered NIPT as a first screen for aneuploidy. NIPT is not a replacement for diagnostic prenatal testing. A positive NIPT result should be confirmed by diagnostic testing (amniocentesis or chorionic villus sampling [CVS]) prior to any irreversible action. The expected benefit of NIPT will be fewer women undergoing secondary invasive diagnostic tests associated with a risk of pregnancy loss.

NIPT is ordered by a healthcare professional. Some genetics centres are counselling patients about this testing option, and some are also offering testing for patients who have been referred because of a high risk indication. All patients should have pre- and post-test counselling to ensure informed decision making and follow-up.

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RISK FLAGS TO CONSIDER TESTING OR GENETIC CONSULTATION

NIPT has been validated for use in women determined to be at high risk of having a fetus with certain aneuploidies (trisomy 13, 18, 21 and X and Y deletion). Consider discussing NIPT as an option for women who:

- Are of advanced maternal age, defined as 35 years of age or older at EDB
- Have an abnormal serum screen (i.e. FTS/IPS/MSS)
- Have a fetal nuchal translucency (NT) measurement of 3.0mm or greater
- Have had a previous pregnancy or child with aneuploidy
- Have fetal congenital anomalies on ultrasound highly suggestive of trisomy 13, 18 or 21
- Have cell markers on ultrasound which are highly suggestive of aneuploidy [Refer to [GEC+KO website](#) 2016].
- Are at risk of carrying a male fetus with an X-linked condition (NIPT could be used for sex determination)

As each prenatal genetics centre has variable referral criteria and practice, abnormalities seen on ultrasound (e.g. congenital anomalies, NT > 3.0mm or other cell markers) should be discussed with [www.mountsinai.on.ca/obgyn](#) to decide whether a referral is appropriate, whether NIPT should be offered first, or if additional testing should be considered.

WHAT DOES THE TEST RESULT MEAN?

Depending on the company, results may be worded as positive or negative, aneuploidy detected, no aneuploidy detected or aneuploidy suspected/false low value or high risk or low risk.

Results typically take approximately 8-10 days.

- If the result is negative, this is reassuring, however NIPT **cannot**:
- detect aneuploidy other than chromosomes 13, 18, 21, X and Y
 - some companies are now adding screening for other trisomies and certain microdeletion syndromes, addition of these rare conditions in the test increases the false positive rate and decreases the positive predictive value
 - completely rule out aneuploidy



Genetics Centres

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Outside Canada

Click here to find up-to-date contact information, referral criteria, requisitions, forms and more for your local **Genetics Centre**.

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GEC+KO

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Updated May 2016

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CALGARY

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Metabolic Disease Clinic Fax: 403-955-3091

Outreach Genetics

Red Deer Phone: (403) 314-5228 or (403) 314-5226 Fax: (403) 346-8830

Medicine Hat Phone: (403) 502-8210 Fax: (403) 528-2250

Camrose Phone: (403) 697-2983 Fax: (403) 697-2999

Lethbridge Phone: (403) 388-6652 Fax: (403) 328-5934

Elnora/Drumheller Phone: (403) 773-3636 Fax: (403) 777-3949

- Contact information
- Requisition
- Referral criteria
- Special instructions

Education Modules

Prenatal and
Preconception Genetics

Adult Genetics

Pediatric Genetics

General Genetic
Counselling

Prenatal and Preconception Genetics

These seminars use a primary care case-based approach to discuss new advances in genomics and how they impact practice.

[Consanguinity \(Nov 2015\)](#)

[Non-Invasive Prenatal Testing \(NIPT\) with microdeletions \(Nov 2015\)](#)

[Prenatal Chromosomal Microarray \(Nov 2015\)](#)

[Expanded carrier screening \(May 2016\)](#)

[Consanguinity \(Nov 2015\)](#)

Additional resources in [GECKO Messenger](#), [GECKO on the run](#) and Point of Care tools in [ethnicity-based screening](#).

Following this session the learner will be able to:

- Refer to their local genetics centre and/or order genetic testing appropriately regarding consanguinity
- Discuss and address patient concerns regarding consanguinity
- Find high quality genomics educational resources appropriate for primary care

[Non-Invasive Prenatal Testing \(NIPT\) with microdeletions \(Nov 2015\)](#)

Additional resources in [GECKO on the run](#) with English and French documents

Following this session the learner will be able to:

- Appropriately refer to their local genetics centre and/or order non-invasive prenatal testing (NIPT)

- Learning modules on various genomic topics
- Case-based learning
- Can be used by educators to facilitate teaching or by individuals motivated to learn more about genomic topics

In-person Seminar Topics

- General:
 - Familial hypercholesterolemia (**2016**)
 - Hereditary hemochromatosis (2013, 2014, 2015)
 - Alzheimer disease (2014, 2015)
 - Multiple sclerosis (2014, 2015)
 - Factor V Leiden (2014)
 - Autism, developmental delay, intellectual disability and Introduction to chromosomal microarray (2013, 2014)
 - Direct-to-Consumer genetic testing (2013, 2014, 2015, **2016**)
- Cancer:
 - Lynch syndrome (2013, 2014, 2015)
 - Hereditary breast and ovarian cancer syndrome (2015, **2016**)
- Cardiogenetics:
 - Hypertrophic cardiomyopathy (2014)
 - Long QT syndrome (**2016**)
- Prenatal & preconception:
 - Non-invasive prenatal testing (NIPT/cfDNA) (2013, 2014)
 - NIPT with microdeletions (2015)
 - Prenatal chromosomal microarray (2015)
 - Expanded carrier screening (**2016**)
 - Consanguinity (2015)

Short-term outcomes

Program Outcome
GEC-KO in common use by Ontario primary care providers.



Summative evaluation

Program Outcome
Provider Outcome

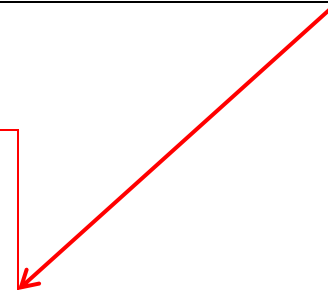
Provider Outcome

- Change in awareness
- Change in knowledge
- Change in confidence in core genetics skills/competencies
- Change in attitudes



Success Criteria

- What will we consider a significant improvement?
- What will we do if we are unsuccessful?



Evaluation

What we have done

In our research, we have evaluated our tools by assessing knowledge, confidence in core genetics skills, vignette management and reflective e-learning

- Carroll JC, Wilson BJ, Allanson J *et al.*, GenetiKit: a randomized controlled trial to enhance delivery of genetics services by family physicians. *Fam Pract* 2011
- Carroll JC *et al.* Efficacy of an educational intervention on family physicians' risk assessment and management of colorectal cancer. *J Community Genet* 2014
- Carroll JC, Grad R, Allanson J *et al.*, The Gene Messenger Impact Project: An innovative Continuing Education Strategy for Primary Care Providers. *JCEHP* 2016

Evaluation

What we are doing

- Seminar evaluation
 - Usefulness of information, relevance
 - Impact on practice; change and improvement
- Participation in research trials which incorporate our resources
- Google and Piwik analytics

Evaluation

What we have yet to do

- Evaluate skills
 - Chart audit for family history completeness
 - Audits of referrals to genetics and other specialist services
 - Audits of appropriateness/completeness of genetic tests ordered by primary care providers
- Repeat our needs assessment survey (summative evaluation)
 - Improved knowledge, awareness, confidence in competencies, attitudes toward genomic medicine



Gaps where additional or modified training experiences would be helpful

- Genomics education needs to be relevant and applicable to the learner's practice (adult learning principles)
- Resources need to be tailored to the learner's needs not necessarily those perceived by the educator
- Ongoing support and resources are needed to support a learner's experience
- Integration into the electronic health record with clinical decision support is needed

Thank you!

Questions?

www.geneticseducation.ca

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