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Genetics Education Canada Knowledge Organization

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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

Arm 1: Primary care providers
Provide care to patients at first contact with the healthcare system, including access, coordination, continuity and comprehensiveness of care as it relates to disorders with a genetic component

Arm 2: Secondary care providers
Provide care to patients who have been referred by primary care or other healthcare providers

Arm 3: Trainees
- Undergraduate medicine
- Postgraduate trainees of various disciplines

Arm 4: The Public

Description of goals, activities conducted and outcomes of program development and delivery will be provided specifically for each arm of the program (Gaff et al., 2007 model)
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

Negotiation and planning

Stakeholder identification and engagement
- Policy makers
- Genetic service providers

Formative Evaluation

Refinement of goals
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

Development
- Topics, content and format determined
- Informed by Needs Assessment/Literature search/Evidence base/Horizon scan of existing quality, evidenced based (when available) resources

Incorporate changes
Review content
- Content experts
- Advisory board working group
- Primary Care Provider representatives

Implementation
- Produce resources
- Disseminate via Website
- Advertise in journals
- Conferences

Formative evaluation

Process evaluation

Improvement of products
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Part I: Hereditary breast and ovarian cancer referral screening tool to identify patients most likely to benefit from referral to genetics

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

Management: With 1 or more positive responses, discuss 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 their 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 (≥2 relatives) and pancreatic cancer
- High risk ethnicity (Ashkenazi Jewish, Israeli) 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 breast 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
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

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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.
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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.4 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, a-globin and/or b-globin.

Thalassemias are due to decreased production of a- or b-globin chains and sickle cell disorders are due to the production of a structurally abnormal b-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

Definition: Consanguinity is defined as a union between two individuals who are related as second cousins or closer. This occurs in consanguineous unions (i.e., marriages between relatives) and is characterized by the sharing of common ancestors. The risk for certain diseases in the offspring of a consanguineous union is estimated based on family history, degree of consanguinity, and geographic population densities. In general, the risk of disease is lower in more distant relatives, whereas diseases in relatives closer than first degree are more common. The risk for a more closely related union is higher and for a more distantly related union is lower. The risk for a first cousin once removed is intermediate, and standard genetic screening should be offered as a general precaution. Detailed genetic counseling can be provided if appropriate based on family history and severing results.

What is Consanguinity?

Definition of the consanguineous union has been a topic of interest for genetic counseling. Consanguinity is defined as a union between two individuals who are related as second cousins or closer. In South Africa, while first and second-degree consanguinity and first and second-degree consanguinity are rare, about 25% of all unions are consanguineous and the first-degree consanguineous for about 5% of all marriages. In Figure 1, the global distribution of consanguinity rates. Reasons for performing such a union can include cultural, social, family, religious, and patrilineal associations associated with health and financial issues. Primary healthcare providers are likely to be exposed to consanguineous unions from those couples who are seeking premarital/prenatal counseling.

Figure 1. Global distribution of consanguinity rates.

What do we need to know about the genetics of consanguinity?

Interested and consanguineous unions often result in consanguinity, although consanguinity is a consanguineous union where one person is consanguineous with one, two, three, or four. The significance of consanguinity?

Risk for offspring where F1/1

Second degree relatives where F1/2

Third degree relatives where F1/3

Fourth degree relatives where F1/4
Non-invasive prenatal testing

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 (~$5000).

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 PREGNATAL 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. cell-free DNA 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 (NIPT)** is a screening test to potentially detect Down syndrome and other congenital abnormalities. NIPT assesses fragments of cell-free DNA (cfDNA) that are circulating in maternal blood. cfDNA can be used to identify chromosomal abnormalities. However, since NIPT is a screening test, it is not a substitute for chorionic villus sampling (CVS) or amniocentesis.

NIPT uses the same technologies as other prenatal screening tests to identify chromosomal abnormalities. However, unlike CVS or amniocentesis, NIPT does not involve obtaining a sample of tissue from the fetus.

NIPT is a safe and minimally invasive test that can be performed at any time during pregnancy. It is often used as an alternative to CVS or amniocentesis for women who are high risk or prefer a non-invasive test.

NIPT results are reported as a likelihood ratio (LR) or a risk ratio (RR). These ratios are used to estimate the probability that a fetus has a genetic abnormality.

NIPT can be performed on a maternal blood sample that is generally safe for the mother. Testing can be conducted as early as 9 weeks pregnant. A detailed explanation is provided prior to blood drawing, including an accurate gestational age, and to eliminate multiple pregnancies.

NIPT is ordered by a healthcare professional. Some genetics services are providing testing for patients who have been referred because of a high risk indication. All patients should have pre- and post-test counseling to ensure informed decision-making and follow-up.

**Red Flags to Consider Testing or Genetic Consultation**

NIPT has been shown to be an effective test for women determined to be at high risk of having a fetus with certain abnormalities (Down, 13, 18, and 21 trisomy). Consider discussing NIPT as an option for women who:

- Are of advanced maternal age, defined as 35 years or older at delivery.
- Have an abnormal serum screen (1ST or 2ND trimester).
- Have a prior affected child or sibling with Down syndrome.
- Have a prior affected child or sibling with 13, 18, or 21 trisomy.
- Have a history of chromosomal abnormalities in other family members.
- Have a history of recurrent miscarriage or infertility.

As with all genetic testing, a referral to a genetic counselor is recommended before testing. This consultation can help to determine whether NIPT is appropriate and can provide additional information about the implications of a positive result.

**What Does the Test Mean?**

Depending on the company, results can be reported as positive or negative. A positive result indicates that the fetus may have a genetic abnormality, but a negative result indicates that the fetus is likely normal. A positive result is further investigated through additional testing.

Results typically take approximately 7-10 days after the sample is received.
Genetics Centres

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

- Contact information
- Requisition
- Referral criteria
- Special instructions
Education Modules

- 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

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In-person Seminar Topics

• General:
  – Familial hypercholesterolemia (2016)
  – Multiple sclerosis (2014, 2015)
  – Factor V Leiden (2014)
  – Autism, developmental delay, intellectual disability and Introduction to chromosomal microarray (2013, 2014)

• Cancer:
  – 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.

Provider Outcome
• Change in awareness
• Change in knowledge
• Change in confidence in core genetics skills/competencies
• Change in attitudes

Summative evaluation
Program Outcome
Provider Outcome

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?

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