Genetics Education Canada Knowledge Organization

Inter-Society Coordinating Committee for Practitioner Education in Genomics Meeting
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**Who**

- Dr. June C. Carroll
  - Co-founder and co-director
  - Professor and Clinician Scientist
  - Family physician

- Dr. Judith E. Allanson
  - Co-founder and co-director
  - Professor
  - Retired clinical geneticist

- Ms. Shawna Morrison
  - Program manager
  - Certified genetic counsellor

**What**

To increase genetics literacy in healthcare professionals and thereby enhance quality of genomic care in order to optimize the health and well-being of Canadians.

**How**

**Funding**

**Development**
Genomic medicine in primary care: 

Needs assessment results

• Brief summary of needs assessments that informed our product development
  • Survey
  • Qualitative
Genomic medicine in primary care: Needs Assessment: Qualitative results

• Wanted
  • Point of care tools
    • Embedded in EMR with clinical decision support
    • Web based
  • Non biased, up to date
  • Connection with genetics
Types of GEC-KO products:
the website www.geneticseducation.ca
Types of GEC-KO products: point of care tools www.geneticseducation.ca

Cardiogenetics

Hypertrophic Cardiomyopathy

Evaluation and Management Tool

Hypertrophic cardiomyopathy (HCM) is a relatively common condition affecting the heart muscle and can present at any age. The evaluation and management of HCM is outlined in the following downloadable point of care tool. The principal role of genetic testing is not to confirm a diagnosis but rather to identify the causative gene in the affected individual and to provide a clinical tool for screening family members at risk of developing the disease. In general, affected individuals and their first degree relatives should be referred to both cardiology and genetics specialists.

Long QT syndrome

Long QT syndrome (LQTS) is one of several inherited heart disorders that can lead to sudden cardiac death (SCD). The downloadable point of care tool contains a brief overview of LQTS and the red flags for how to identify LQTS and the individuals who would most likely benefit from referral to genetics and a cardiac arrhythmia specialist.
Types of GEC-KO products:

point of care tools

www.geneticseducation.ca
Types of GEC-KO products: point of care tools www.geneticseducation.ca

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

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-Proalla et al 2009), which was designed for use in primary care settings and demonstrated an overall sensitivity of 97.0% and a specificity of 93.0% 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.


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Types of GEC-KO products:

GEC-KO on the run www.geneticseducation.ca

Familial hypercholesterolemia

WHAT IS FAMILIAL HYPERCHOLESTEROLEMIA?

Familial hypercholesterolemia (FH) is a common autosomal dominant genetic condition where the uptake of low-density lipoprotein (LDL-C) into cells is either decreased or impaired. Most cases (80-90%) of FH are caused by mutations in the LDL receptor gene (LDLR). This results in the body’s inability to cope with high levels of LDL-C, which can lead to a 20-fold increase in premature coronary artery disease (CAD) and death. Early diagnosis and treatment can normalise the cholesterol homeostasis. Key factors of FH are elevated LDL-C (150 mg/dL), with additional features such as early onset CVD (<35 years in men, <45 years in women), atherosclerotic plaques, in the arteries, and family history of early onset CAD or hypercholesterolemia requiring treatment. Genetic screening of family members with LDL-C levels or genetic testing for the familial gene mutation when possible, allows for early identification and treatment of at-risk individuals, with statins as first-line treatment.

Table 1. Clinical features of familial hypercholesterolemia in heterozygote (HeFH) and homozygote (HoFH)

<table>
<thead>
<tr>
<th>Clinical features</th>
<th>HeFH</th>
<th>HoFH</th>
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<tbody>
<tr>
<td>LDL-C levels</td>
<td>5-7 mg/dL</td>
<td>&lt;3 mg/dL</td>
</tr>
<tr>
<td>Cardiovascular disease symptoms</td>
<td>&gt;60 years of age in men</td>
<td>&lt;5 years of age in children</td>
</tr>
<tr>
<td>Physical findings</td>
<td>Cholesterol deposits in the tendons (xanthomas) and/or around the eyes (xanthelasma)</td>
<td>Arteriosclerotic plaques, grey or blue opaque ring in the corneal margin (xanthoma)</td>
</tr>
<tr>
<td>Family history</td>
<td>Early onset CVD</td>
<td>No family history</td>
</tr>
</tbody>
</table>

Download the PDF here. Download the point of care tool to easily identify patients most likely to benefit from referral to a specialist or for a more comprehensive review. The GEC-KO Messengers containing more on diagnosis, screening, and management are here. Access an education module with case-based learning here.
Types of GEC-KO products:

GEC-KO on the run www.geneticseducation.ca
Types of GEC-KO products:

**GEC-KO Messenger** www.geneticseducation.ca
Types of GEC-KO products:

GEC-KO Messenger www.geneticseducation.ca
Types of GEC-KO products:

Contact to centres [www.geneticseducation.ca](http://www.geneticseducation.ca)
Types of GEC-KO products:

Contact to centres www.geneticseducation.ca

- Contact information
- Requisition
- Referral criteria
- Special instructions
Types of GEC-KO products:

In person seminars www.geneticseducation.ca

- 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
Types of GEC-KO products:

In person seminars www.geneticseducation.ca

• General:
  • Familial hypercholesterolemia (2016)
  • Alzheimer disease (2014, 2015)
  • 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)
Types of GEC-KO products:

In person seminars www.geneticseducation.ca

• Time:
  • 60-90 minutes
  • ~25% interactive

• Several topics in a session

• Format:
  • Case-based
  • Basic genetics (inheritance, prevalence)
  • Red flags for genetic referral and/or testing
  • Genetic test results (positive, true negative, uninformative, VUS)
  • Screening and surveillance
  • Pearls
Evaluation

• RCT of *GEC-KO Messengers (GM)* showed significant increase in:
  • appropriate referral to genetics
  • self reported confidence in core genetic competencies
    • *Carroll et al Family Practice 2011*

• Email “push” of GM to members of the College of Family Physicians of Canada
  • Used Information Assessment Method (cognitive impact, relevance, intended use for a patient, expected health benefits)
  • 73% indicated practice would be improved after reading GMs
  • Of those who rated a GM relevant, 94% would apply it to at least 1 patient and 70% expected health benefits
    • *Carroll et al JCEHP 2016*

• Seminar evaluation
  • Good
  • Too much content
Evaluation

website analytics [www.geneticseducation.ca](http://www.geneticseducation.ca)

Users

<table>
<thead>
<tr>
<th></th>
<th>Users</th>
<th>Page views</th>
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<tr>
<td>Int1: Jun 1 2015 to Jan 1 2016</td>
<td>2,117</td>
<td>5,478</td>
</tr>
<tr>
<td>Int2: Jun 1 2016 to Jan 1 2017</td>
<td>5,478</td>
<td>9,295</td>
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Evaluation website analytics [www.geneticseducation.ca](http://www.geneticseducation.ca)

User location

<table>
<thead>
<tr>
<th>Top 5 Countries</th>
<th>Int 1: June 1 2015 - Jan 1 2016</th>
<th>Int 2: June 1 2016 - Jan 1 2017</th>
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<tr>
<td>1</td>
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<tr>
<td>5</td>
<td>France</td>
<td>Australia</td>
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</table>
Evaluation
website analytics  www.geneticseducation.ca
User behaviour

Website section (% of total page views, 9,295)

1. Educational Resources (53%, N = 4,943)
   - GEC•KO on the run (3,738)
   - Non-invasive prenatal testing (NIPT) (2,928)
   - Hereditary breast and ovarian cancer syndrome (210)

2. Genetics Centres (10%, N=924)
   - GEC•KO Messenger (463)
   - Consanguinity (194)
   - Hereditary breast and ovarian cancer syndrome (70)

3. Point of Care (POC) tools (7%, N=635)
   - Family history (166)
   - Hereditary cancer (166)
What works

• Be evidence-based and brief, get to the point quickly
• Keep resources up-to-date and locally relevant
• Limit barriers to information access e.g. no sign in
• Provide resources for point of care
• Integrate into existing education venues
• Use interactive workshop format
• Engage and listen to stakeholders
  • Be flexible, responsive, continuously evolve
  • Meet clinical needs and questions of stakeholders
Challenges

• Engaging with primary care providers
• Implementation into practice
• Evaluation
• New format

www.geneticseducation.ca

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