COGR
Canadian Open Genetics Repository

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BRCA1/2 Related Breast Cancer Risk
Hereditary Breast and Ovarian Cancer Genetic Testing in Ontario

Database

- CHEO
- Mount Sinai
- Kingston
- North York
- Credit Valley
- London
- Hamilton
Major barriers

1) Evolving technologies - vast amount of data
2) Reporting and communication (more complicated) – time consuming
3) Biological complexity - clinical heterogeneity
4) Differences in rules for classifying variants
5) Rare mutations; rate of newly identified variants
6) Poorly annotated variant databases
7) Variant classification changes over time
Status quo not an option...

Genetic data scattered across institutions and locked behind hospital firewalls

Protocols, procedures vary widely (e.g. differences in hospital information systems, variant classifications and reporting)

Errors, inconsistencies too common
- a disservice to society
Canadian Open Genetics Repository

- To create Canada’s unified, open-access, clinical-grade genetic database using a commonly shared platform

- Designed to hold all types of information related to human gene variants and their relationship to rare and common diseases

- Also, create and disseminate variant assessment procedures to each laboratory to help standardize clinical classification
This work was funded by the Government of Canada through Genome Canada and the Ontario Genomics Institute (OGI-070)
Our major goal...

Moving collaboration to a whole new level for research + patient care

Lab directors - with clinicians, GCs, bioinformaticians, scientific community

To greatly expand quality, scale, scope of our work
Participating Laboratories
Not just a Canadian challenge...

Major projects funded in the US

Building on existing ClinVar database

Wide acceptance among US labs

Support for our project - inc. GenelInsight®
Adds strong resources to Cdn project
An International Effort
Three main aims...

Design variant assessment procedures

Extract and transfer data

Set up access and dissemination
Role of bioinformatics and IT...

Build system for variant interpretation

Define ontologies, structured phenotype data

Extract de-identified variant knowledge

Facilitate public access
A Canadian GeneInsight instance

- No limit to number of users
- Ability to share data within other labs using the system
- Each lab will need to make their own decision on whether they are comfortable sharing data with the Canadian installation
- Incent labs to share, in a share and share alike fashion
- Pulls ClinVar data into GeneInsight so it becomes accessible through the system
- supports pushing data into ClinVar for labs who are open to releasing data to the public
## GeneInsight - DNA Variant Knowledgebase

### Variants (753)

**Gene Name/Symbol contains BRCA1**

<table>
<thead>
<tr>
<th>Gene</th>
<th>DNA</th>
<th>AA</th>
<th>**</th>
<th>Region</th>
<th>Cat (Dis)</th>
<th>Cat Date</th>
<th>Ret</th>
<th>Fam</th>
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<tr>
<td>BRCA1</td>
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</table>
Data Release: 1
Upload From Lab
Integration with LMS is Possible

‘Pull’ of record subsets

LMS or EMR

Lab Store

Auto-Upload

Auto-Upload

Variant

SiteA.GenelInsight
Direct Database Query
‘Pull’ of Variant Information

- Custom Script
- SOAP API Interface
- SiteA.GeneInsight
- Lab Store
- Computational analysis
Data Release: 2
Internal to Consortium

COGR ‘Consortium’

COGR.GeneInsight
(consensus interpretation)

SiteA.GeneInsight

SiteB.GeneInsight

Variant

Lab Store

Project
WWW Portal
Public
DB

Lab Store
Data Release: 3
To Public / External
Variant sharing in GenelInsight
Variant sharing in GenelInsight

DSC2  c.1787C>T (p.Ala596Val)
Interpretation Unk Sig  Found In 2 Reports, 2 Families
APPROVED

Assessments (1) ▶

Frequency (2) ▶

Seq Alignments (2) ▶

References (2) ▶

Networked Labs ▶

Warning: The Network Laboratories section below displays information for outside laboratories or content providers. GenelInsight has not validated this content. It is important that you review and validate this content before using it for any purpose. GenelInsight provides access to these external information sources for convenience only; the inclusion of a laboratory or content provider is not intended to be, and should not be taken as, an endorsement or validation by GenelInsight of any content, product or service that may be available. Terms of use may be different. Any questions regarding the information provided should be directed to the networked laboratory or content provider.
Variant sharing in GeneInsight
VariantWire
Collaboration and sharing of knowledge will enable more accurate interpretation and improve patient care.
This work was funded by the Government of Canada through Genome Canada and the Ontario Genomics Institute (OGI-070)