Updates from the Canadian Open Genetics Repository (COGR)

Marina Wang [1], Shana White [2], Samantha Baxter [2], Michael Oates [2], Chet Graham [2], Matthew S Lebo [2,3], Jordan Lerner-Ellis[1], and the COGR Working Groups

[1] Mt Sinai Hospital, University of Toronto, Ontario Institute for Cancer Research ; [2] Laboratory for Molecular Medicine, PPM; [3]Brigham and Woman's Hospital, Harvard Medical School

Background

Individual laboratory knowledge and soiled data sets impede our knowledge of variants and prevents clinicians from receiving the most accurate interpretations for variants found in their patients. This in turn may prevent patients from receiving the most appropriate care.

The Canadian Open Genetics Repository is a collaborative effort to create a unified, open-access, clinical-grade database of variants reported by medical diagnostics laboratories across Canada. Utilizing GeneInsight® as a common platform, labs will collect, store and share variant information in real time.

As clinical laboratories adopt modern genomics technologies, the need for this type of collaborative framework is increasingly important.

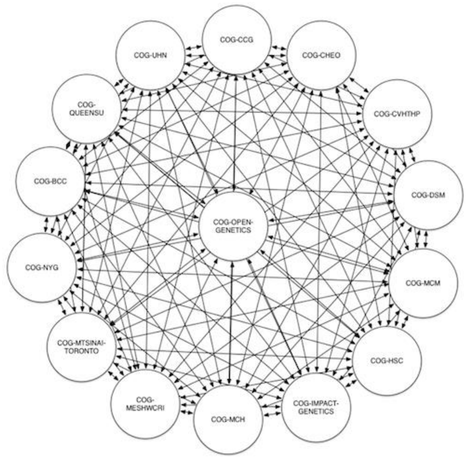
Participating Centers with GeneInsight Instance

Organization	Variant Upload Status	Variant Sharing Status
Alberta Children's Hospital, Calgary AB	Uploading	
British Columbia Cancer Agency, Vancouver BC	Uploading	
Children's & Women's Health Centre of BC, Vancouver BC	Pending	
Children's Hospital of Eastern Ontario, Ottawa ON	Uploaded	Sharing*
Credit Valley Hospital, Trillium Health Centre, Mississauga ON	Uploaded	Sharing
Impact Genetics Inc., Bowmanville ON	Pending	
McGill University Health Complex, Montréal QC	Uploaded	Pending
McLaughlin Centre, SickKids Hospital, Toronto ON	Uploading	
McMaster University (Hamilton Health Sciences), Hamilton ON	Pending	
Memorial Health University Medical Center, St. John's NL	Pending	
Mount Sinai Hospital, Toronto ON	Uploaded	Sharing*
North York General Hospital, Toronto ON	Pending	
Queen's University, Kingston General Hospital, Kingston ON	Uploaded	Pending
Women's College Hospital, Toronto ON	Uploaded	Sharing
University of Alberta, Dept of Medical Genetics, Edmonton AB	Uploading	
University Health Network, Toronto, ON	Uploaded	Sharing
University of Manitoba, Regional Health Authority, Winnipeg MB	Pending	
University of Montreal, Sainte-Justine Hospital, Montréal QC	Uploading	
Western University, University Hospital, London, ON	Pending	

* Sharing with VariantWire

Data Sharing Model

Our progress thus far has highlighted the critical need for robust and sustainable IT infrastructure. Our goal is that continued use of a common variant assessment tool and real-time variant sharing will increase standardization and discussion.



Aims & Platform

Aim 1. Design freely available and consistent variant assessment procedures.

- Individual variant data will be transformed into a unified format, eliminating discrepancies, omissions, and duplications.

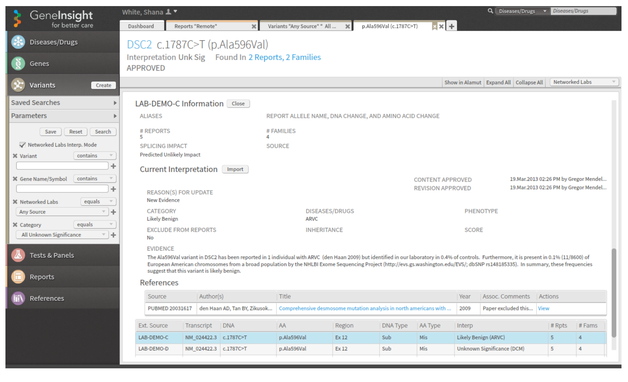
Aim 2. Data extraction and transfer.

- The project team will devise and optimize procedures to support the extraction of disease, gene and variant data currently held within participating laboratories.

Aim 3. Data access and dissemination.

- Methods will be developed such that accurate and readily accessible data will be presented to different stakeholder groups in appropriate summary levels. Individual laboratories see their variants with specific detail while the diagnostic community views a more general summary that is mindful of privacy and confidentiality.

COGR will provide an instance of the GeneInsight platform to all participating labs.



Current Variant Sharing

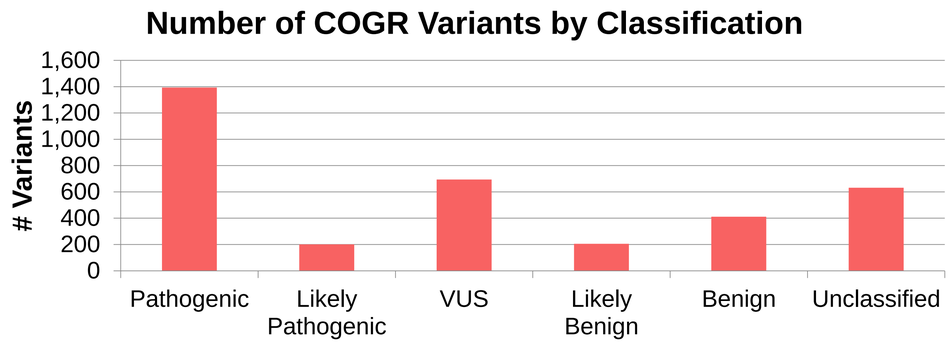
Participating laboratories are currently sharing:

Total Number of Variants: 3802

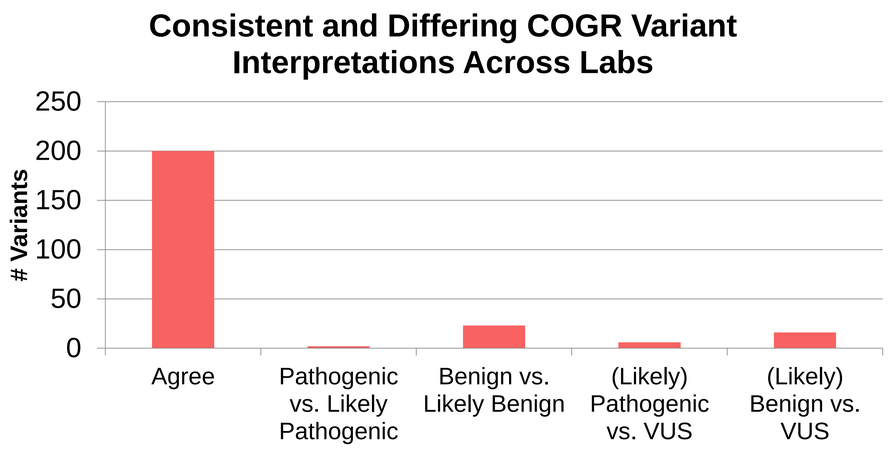
Total Number of Genes: 56

Total Number of Diseases: 13

Disease	# of Variants Linked to Disease
Breast & Ovarian	1130
Unspecified cancer	252
Hereditary Cardiomyopathy	107
Lynch syndrome	84
Phenylketonuria	83
β-Thalassemia	44
A1AD	30
Familial adenomatous polyposis	30
Deafness	21
Cystic Fibrosis	19
MUTYH-associated polyposis	7
Sickle Cell	2



Unclassified variants are under review, pending classification.



Over 3,000 variants are being shared

- 391 have been reported by 2 different labs
- 50 have been seen by 3 different labs

Future Directions

The COGR project is positioned to help remove some of the chief obstacles to advancements in personalized healthcare including the lack of standardized resources and protocols for interpreting the ever-increasing volumes of patient data being generated by clinical labs.

Continuing initiatives include:

- Automatic or semi-automatic the variant upload status for sites to facilitate the maintenance of updated variant information for all sites
- Make consensus variant interpretations available to stakeholder groups with different but appropriate levels of summary.
- Continue to collaborate with other international data-sharing efforts including ClinVar.

In summary, the COGR serves as a focal point for the collaboration of Canadian laboratories with themselves and other countries in the development of tools and methods that take full advantage of laboratory data in diagnosing, managing and treating genetic diseases. Furthermore, as more laboratories share data, knowledge will improve and ultimately lead to better patient care.

Canadian Open Genetics Repository
Email: cogr@opengenetics.ca
Web: **http://opengenetics.ca**