Updates from the Canadian Open Genetics Repository (COGR)

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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.

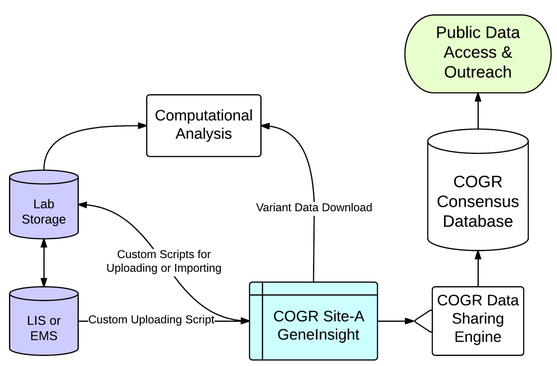
Participating Centers with GeneInsight Instance

Organization	Upload Status	Sharing status
Alberta Children's Hospital, Calgary AB	Uploading	-
British Columbia Cancer Agency, Vancouver BC	Pending	-
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	-
SickKids Hospital and McLaughlin Centre, Toronto ON	Uploaded	Sharing
Hamilton Health Sciences, McMaster University, Hamilton ON	Pending	-
Memorial Health University Medical Center, St. John's NL	Pending	-
Mount Sinai Hospital, University of Toronto, Toronto ON	Uploaded	Sharing*
North York General Hospital, Toronto ON	Pending	-
Ontario Institute of Cancer Research (OICR), Toronto ON	Uploading	-
Kingston General Hospital, Queen's University, Kingston ON	Uploaded	Sharing
Dept of Medical Genetics, University of Alberta, Edmonton AB	Uploading	-
Regional Health Authority, University of Manitoba, Winnipeg MB	Pending	-
Sainte-Justine Hospital, University of Montreal, Montréal QC	Pending	-
University Hospital, Western University, London ON	Pending	-
Women's College Hospital, University of Toronto, Toronto ON	Uploaded	Sharing*

* Sharing with VariantWire

Data Sharing Model

COGR is providing an instance of the GeneInsight platform to all participating labs. The COGR data sharing model allows real-time variant sharing between labs. Ultimately a database of variant interpretations will be created for public access.



Aims & Platform

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

- A free version of the Variant Assessment Tool has been made available on the COGR website.

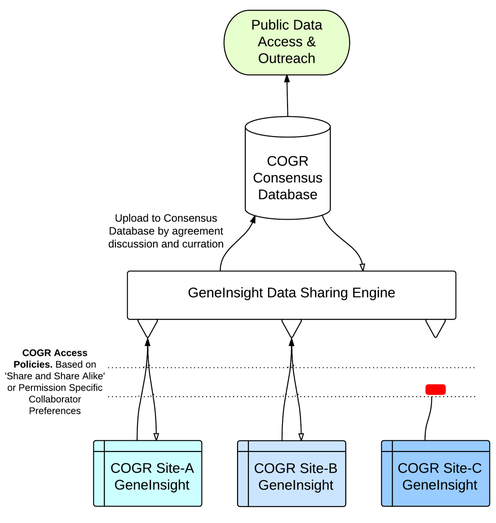
Aim 2. Data extraction and transfer.

- The project team is currently supporting the extraction of variant data currently held within participating laboratories.

Aim 3. Data access and dissemination.

- Methods are being developed such that accurate and readily accessible data will be made public.

Below is a schematic of the COGR project. Note that in this example Site-C has chosen not to share its variant information.



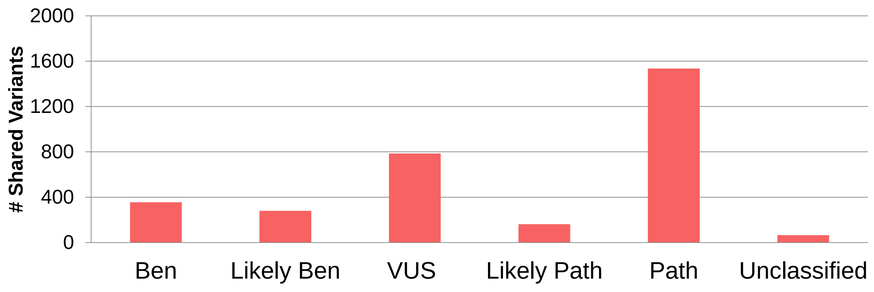
Current Variant Sharing

Currently Shared:

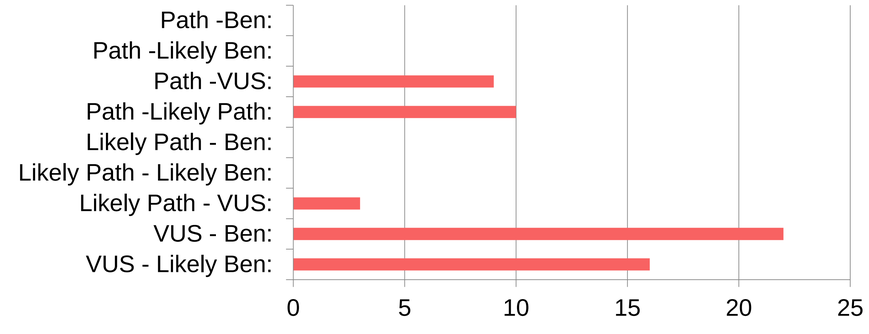
Total Shared Variants:
3280
Total Unique Variants:
2819
Total Genes:
24
Total Number of Diseases:
11

Disease	# of Variants
Hereditary Breast and Ovarian Cancer	2149
Hypertrophic Cardiomyopathy	581
Lynch Syndrome	215
Familial Adenomatous Polyposis	110
Cystic Fibrosis	97
Rett Syndrome	50
MUTYH-Associated Polyposis	33
Autosomal Recessive Non-syndromic Deafness	17
Pompe Disease	14
Clouston syndrome	7
α-1-antitrypsin deficiency	4
Other	3

Number of Shared COGR Variants by Classification



Number of Discrepant COGR Variants



# of Variants Seen By:

Only 1 Lab	2339
2 Labs	250
3 Labs	68
4 Labs	21
5 Labs	3

Total Agreements:
237
Total Disagreements:
96

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:

- Automation or semi-automation of the variant upload process 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
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