

Canadian Open Genetics Repository

October 2015



COGR
Canadian Open
Genetics Repository

Conflict of Interest

Shana White is an employee of Partners HealthCare which has a financial investment in GeneInsight Inc.

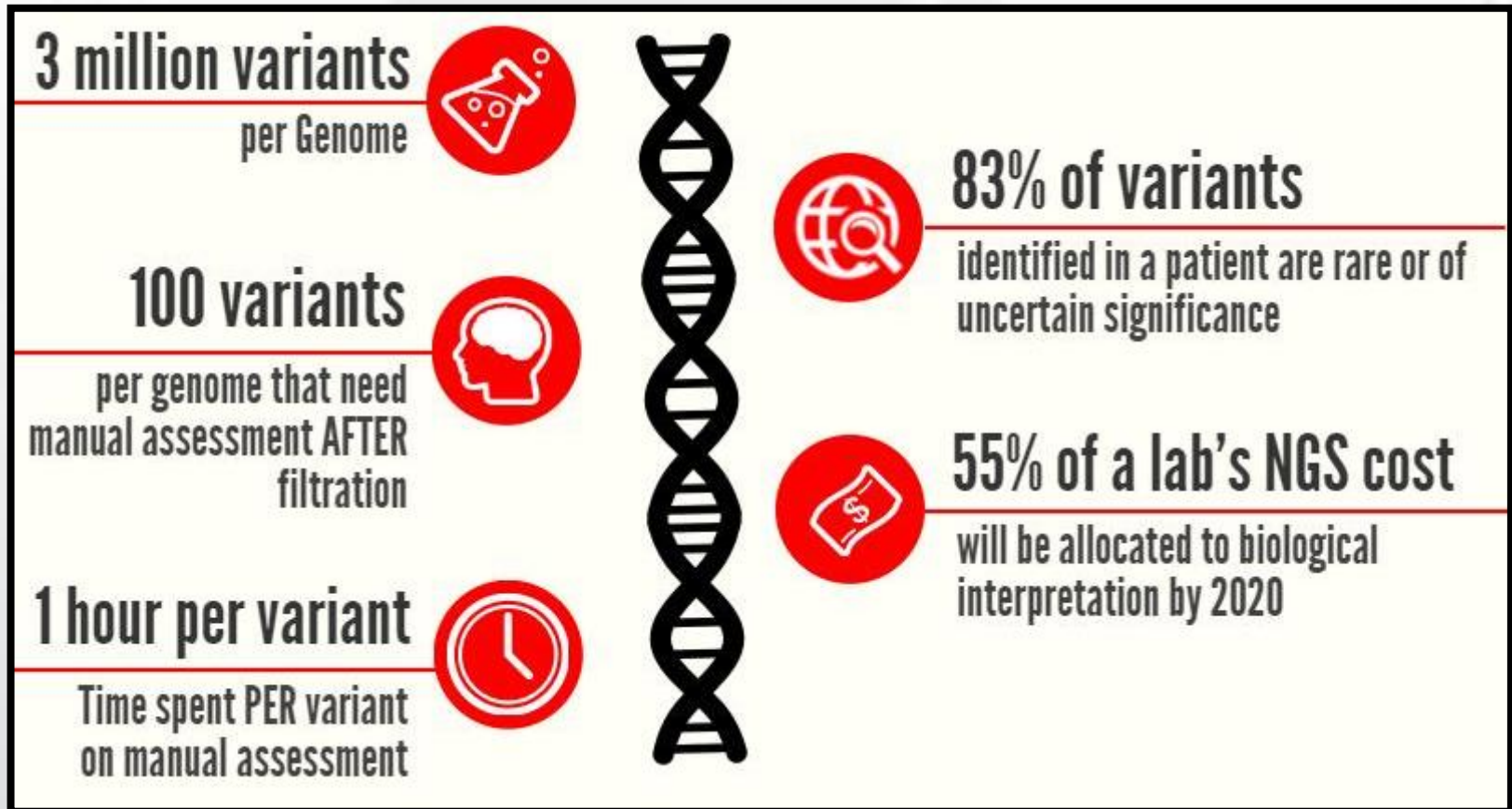
Agenda

1. What is the Canadian Open Genetics Repository?
2. Aims
3. Status
4. BRCA1/2 as proof-of-concept
5. How can you support this initiative?

Overview

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

Why is this needed?



Overview

Standardize variant assessment procedures

Create variant classification **consensus**

Disseminate results to a large public repository

Overview

- Grant funded by Genome Canada
- Collaborative effort for the collection, sharing and analysis of variants reported by medical diagnostic laboratories across Canada
- Collaborate with other global initiatives including ClinVar, BRCA Challenge, Human Variome Project

Aims

1. Design of variant assessment procedures
2. Data extraction and transfer
3. Data access and dissemination

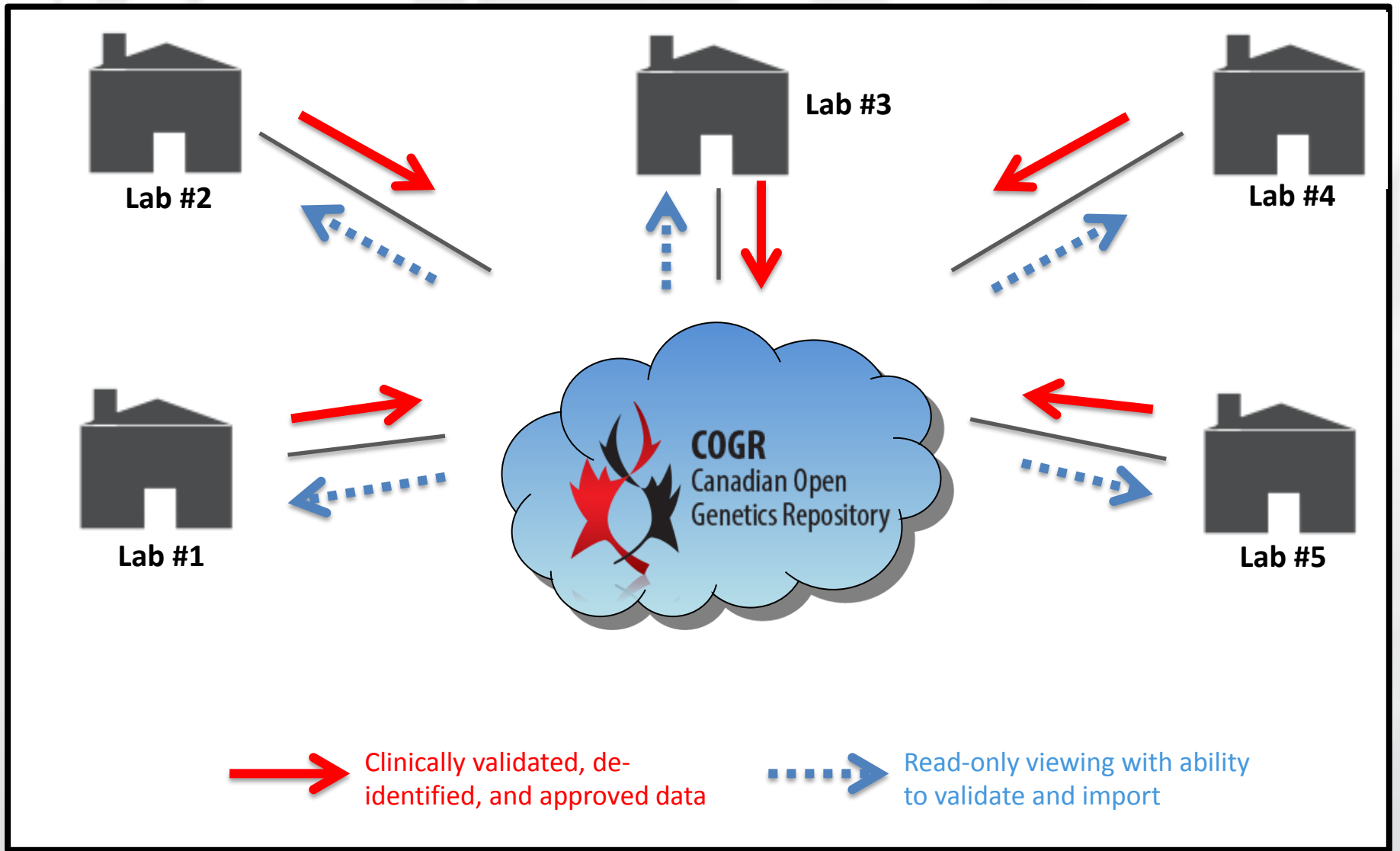
Status of Aims

1. Design of variant assessment procedures
 - *Complete but ongoing*
2. Data extraction and transfer
 - *Ongoing*
3. Data access and dissemination
 - *Lab repository created*
 - *Public repository winter 2016*

What is being shared

- Diseases
- Genes
- Variants and Interpretations

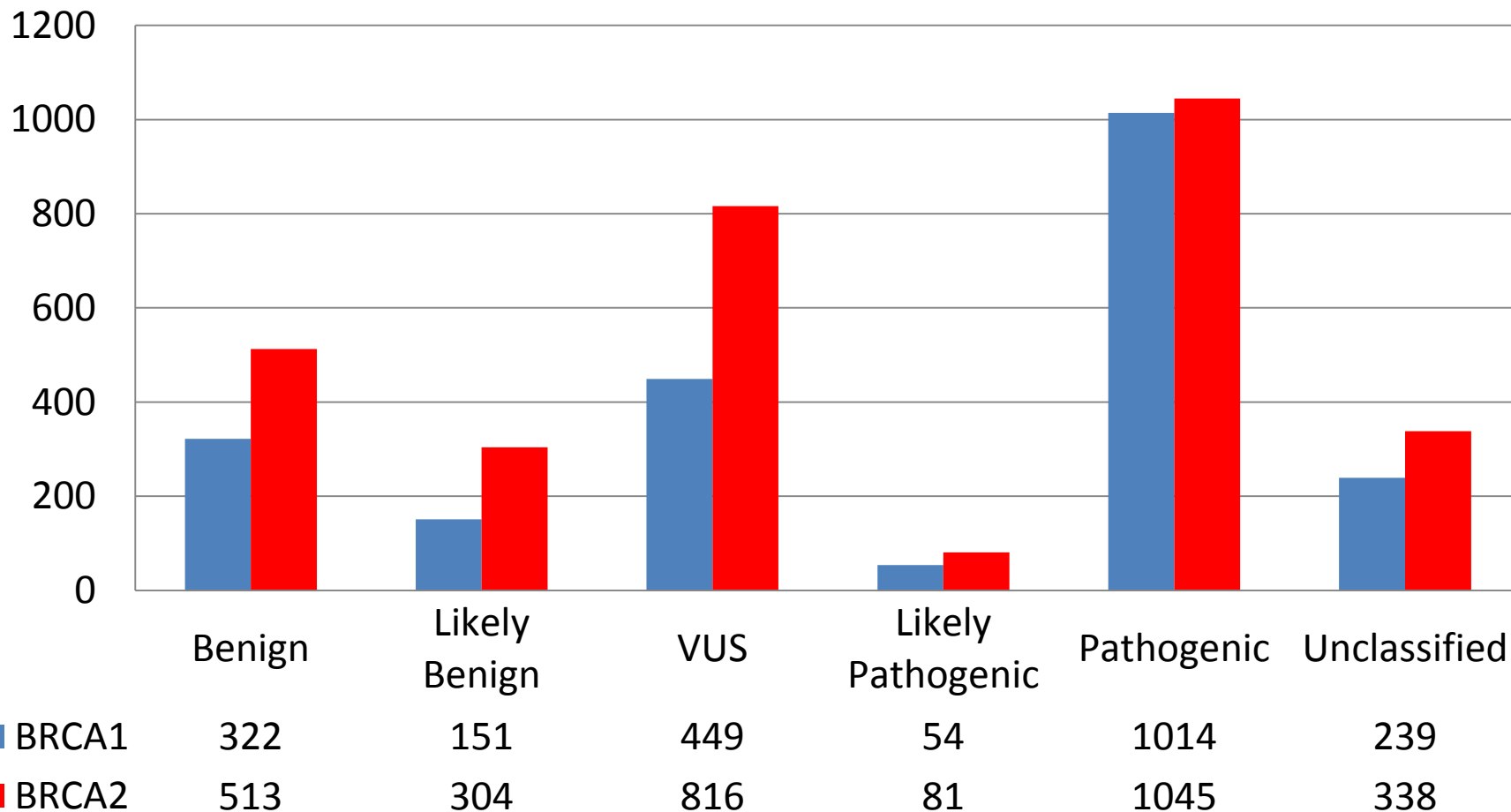
De-identified, clinically validated data in real-time.



Organization	Upload Status	Sharing Status
Alberta Children's Hospital, Calgary AB	Uploaded	Sharing
Atlantic Cancer Research Institute	Pending	-
British Columbia Cancer Agency, Vancouver BC	Uploaded	-
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
Dept of Medical Genetics, University of Alberta, Edmonton AB	Uploading	-
Hamilton Health Sciences, McMaster University, Hamilton ON	Uploading	-
Impact Genetics Inc., Bowmanville ON	Pending	-
Izaak Walton Killam Health Centre, Dalhousie University, Halifax NS	Pending	-
Kingston General Hospital, Queen's University, Kingston ON	Uploaded	Sharing
McGill University Health Complex, Montréal QC	Uploaded	-
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	Uploaded	Sharing
Ontario Institute of Cancer Research (OICR), Toronto ON	Uploaded	Sharing
Regional Health Authority, University of Manitoba, Winnipeg MB	Uploading	-
Sainte-Justine Hospital, University of Montreal, Montréal QC	Uploading	-
SickKids Hospital and McLaughlin Centre, Toronto ON	Uploaded	Sharing
University Hospital, Western University, London ON	Pending	-
Women's College Hospital, University of Toronto, Toronto ON	Uploaded	Sharing*



COGR BRCA Variant Numbers To Date



Variant Classification	Number of Variants
Pathogenic	2059
Likely Pathogenic	135
Variant of Unknown Significance	1265
Likely Benign	455
Benign	835
Unclassified	577
<i>Consistent Interpretations</i>	
Agree	532
Pathogenic or Likely Pathogenic	42
Benign or Likely Benign	130
Total Consistent Interpretations	704
<i>Different Interpretations</i>	
(Likely) Pathogenic vs. VUS	48
(Likely) Benign vs. VUS	246
(Likely) Pathogenic vs. (Likely) Benign	16
Total Different Interpretations	653

How can you support this initiative?

- Ask your lab if they are part of COGR
- Refer to publicly available website when it is released for consensus interpretations and information
- Encourage global data sharing initiatives

Acknowledgements

Thank yous



COGR
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For more information:

OpenGenetics.ca



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