8:30 am - Auditorium
Exploring the Key Issues

12:00 pm - Gallery
Lunch

12:45 pm - Auditorium
Reaching Consensus in the UK and Belgium

2:00 pm – Gallery
Breakout Session to Reach Consensus

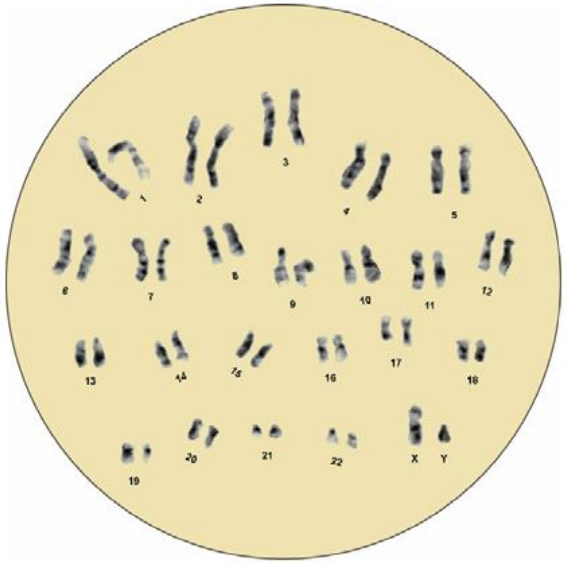
4:00 pm - Auditorium
Group Presentations

5:10 pm – 13th Floor
Wine and Cheese

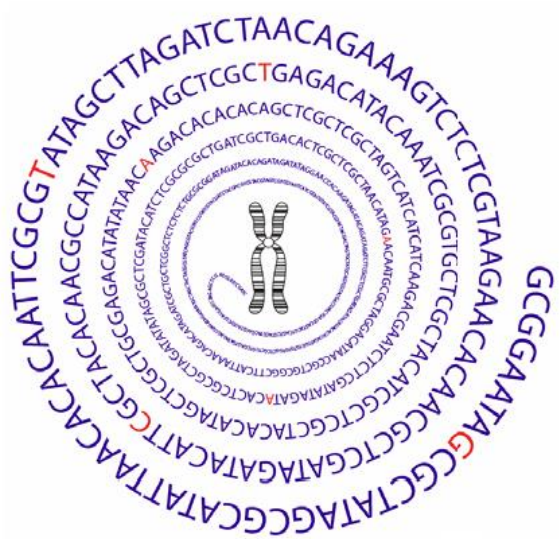
McLaughlin Centre
UNIVERSITY OF TORONTO

Why are you here?

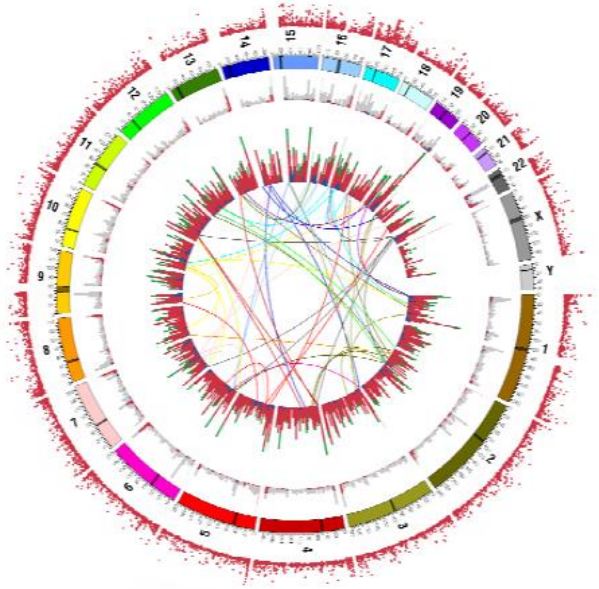
Relax, contemplate, deliberate on prenatal genomics



Chromosomes and alterations



SNPs and gene mutations



Unified genetic variation maps of genome i.e. genomics Nature 2006;, Nature 2010

Deliverables?

Good Canadian common sense

(and maybe a paper and some funding ideas)

UNIVERSITY OF TORONTO
FACULTY OF MEDICINE

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Advancing Genomic Medicine through Research and Education

News

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THE ROYAL SOCIETY OF CANADA ANNOUNCES 2014 MCLAUGHLIN MEDALLIST

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PRENATAL GENOMIC DIAGNOSTICS SEMINARS 2014

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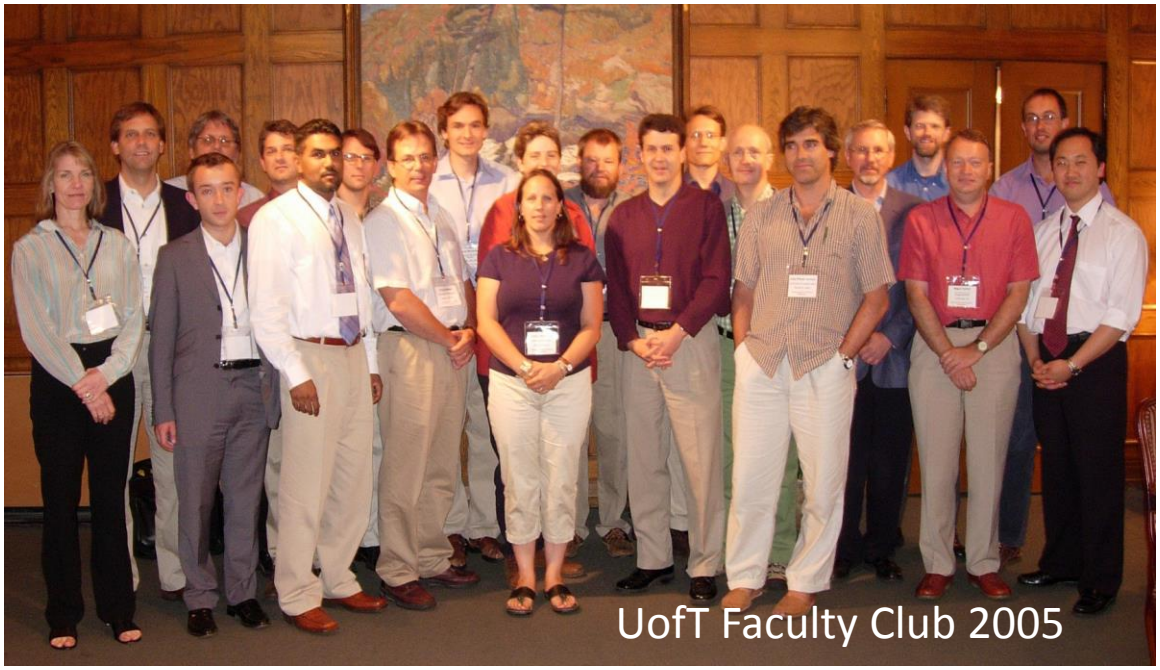
More Announcements

Key McLaughlin Centre Meetings



- Genome Structural Variation Symposium, July 2005
- Micoarray Use in Canadian Clinical Cytogenetics Laboratories, July 2010
- Genetic risk Factors for Autism: Translating Discoveries into Diagnostics, September 2010
- **Canadian Prenatal Genomics Microarray and Sequencing Symposium, October 2014**

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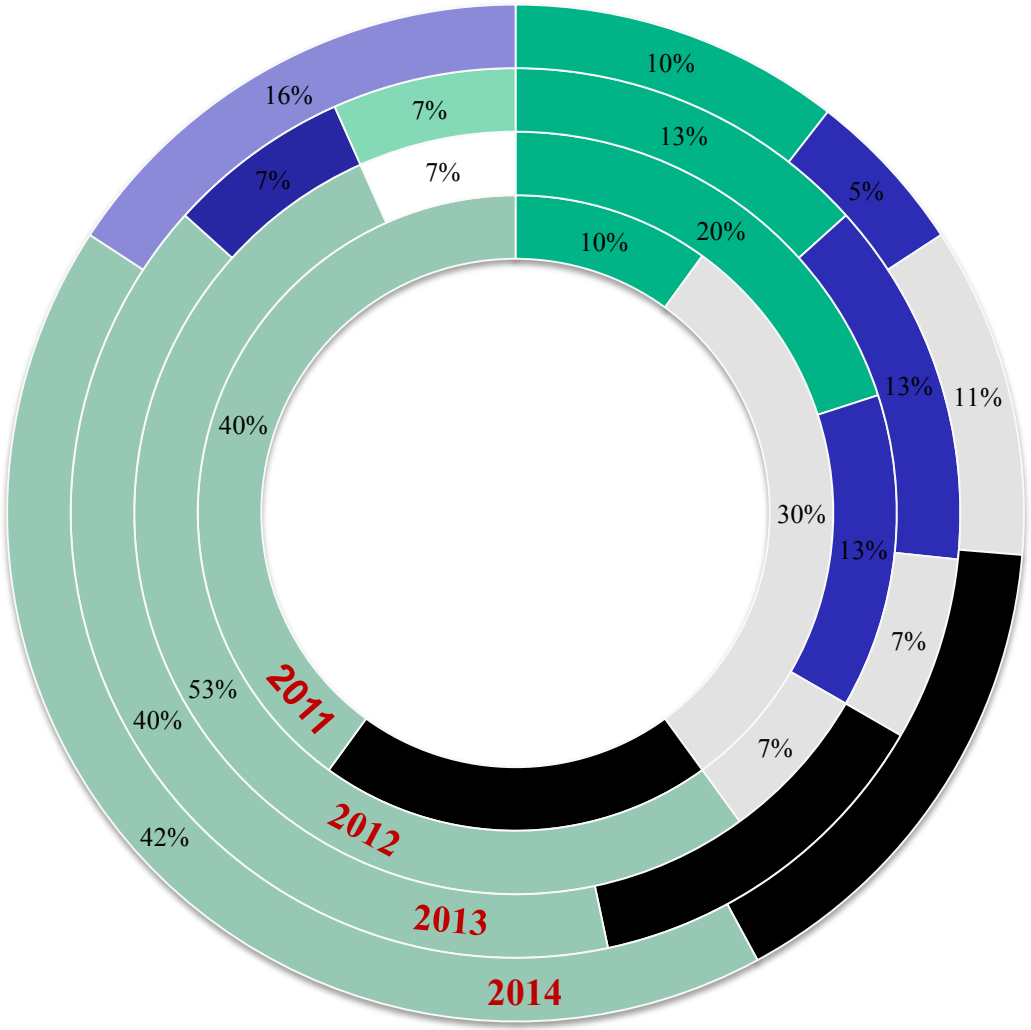
UNIVERSITY OF TORONTO
Advancing genomic medicine through research and education

McLaughlin Center Genomic Medicine Peer-Reviewed Accelerator Grants by Lead Institutions 2011-14



Year	Total Funded
2011	10
2012	15
2013	15
2014	19
	Total: 59

Institutions receive funding

- Centre for Addiction and Mental Health
- Mount Sinai Hospital
- University of Toronto
- University Health Network
- The Hospital for Sick Children
- Public Health Ontario
- Women's College Hospital
- St. Michael's Hospital
- Sunnybrook Health Sciences Centre

Launch of the Personal Genome Project

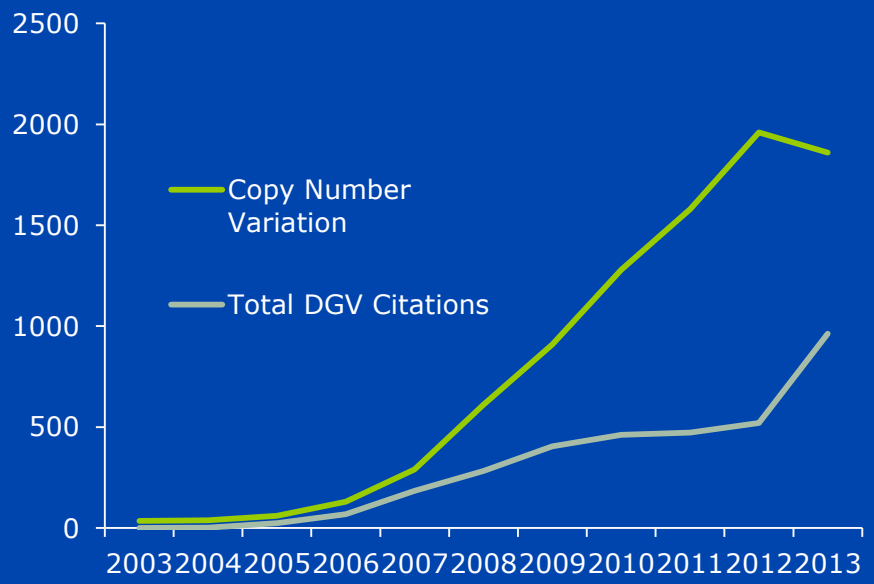
Globe and Mail Saturday December 8th, 2012

"Canada is the only G8 country with no law against genetic discrimination"

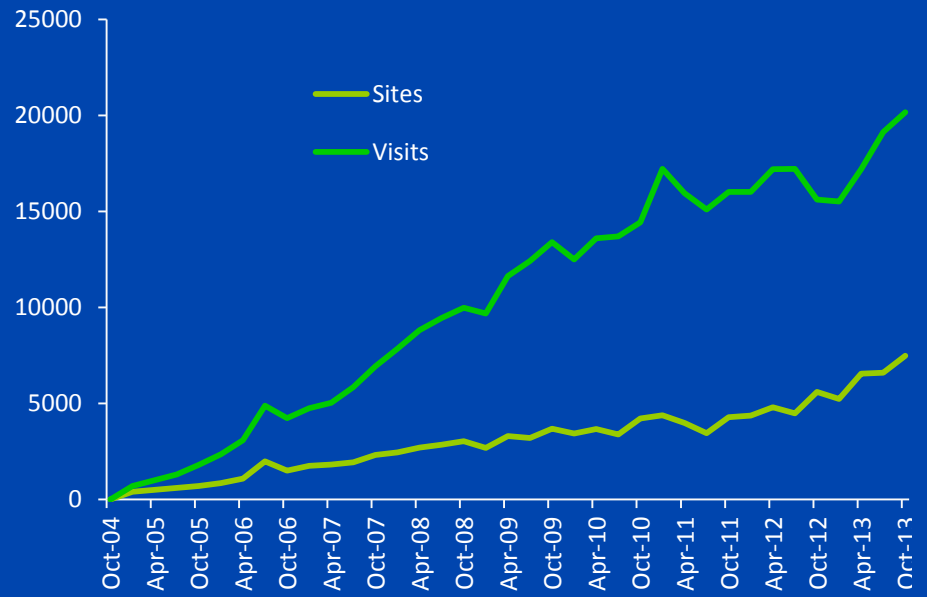
- PGP-C is a joint venture between the University of Toronto McLaughlin Centre, The Centre for Applied Genomics at the Hospital for Sick Children, Medcan Clinic and many other important partners, most notably the USA-PGP.

Database of Genomic Variants

Post 2004 Publication/Citations



Visits to The Database of Genomic Variants



DGV is acknowledged >400 times per year in the peer-reviewed literature.

Nucleic Acids Research, 2013, 1–7
doi:10.1093/nar/gkt958

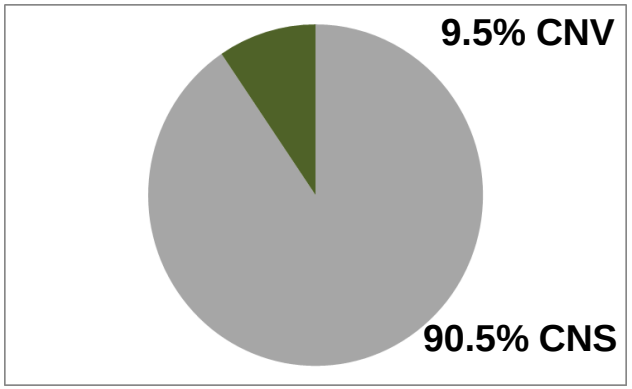
The database of genomic variants: a curated collection of structural variation in the human genome

Jeffrey R. MacDonald[1], Robert Ziman[1], Ryan K. C. Yuen[1], Lars Feuk[2,*] and Stephen W. Scherer[1,3,*]

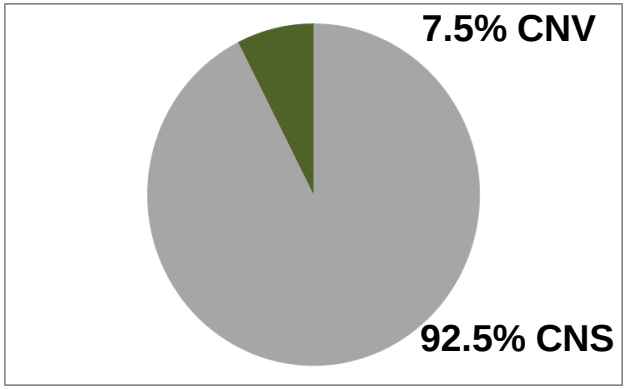
[1]The Centre for Applied Genomics, Peter Gilgan Centre for Research and Learning, The Hospital for Sick Children, 686 Bay Street, Toronto, Ontario M5G 0A4, Canada, [2]Department of Immunology, Genetics and Pathology, Science for Life Laboratory, Uppsala University, Uppsala SE-751 08, Sweden and [3]Department of Molecular Genetics, University of Toronto, Toronto, Ontario M5S 1A8, Canada

A Chromosome Imbalance Map of the Human Genome

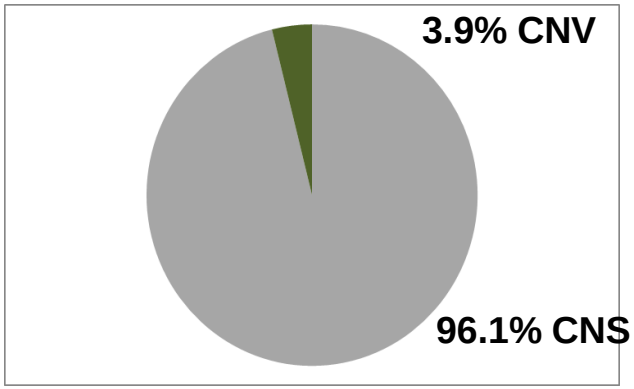
All variants

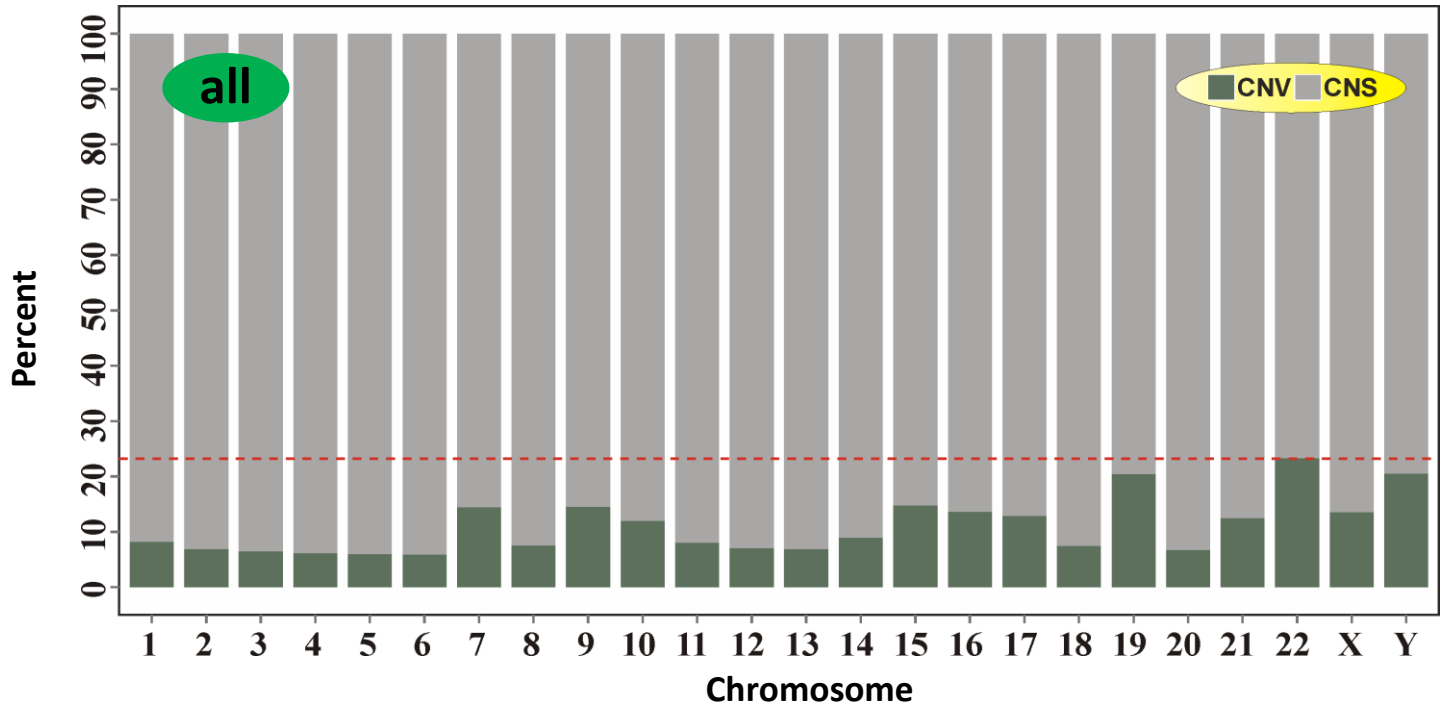


Loss



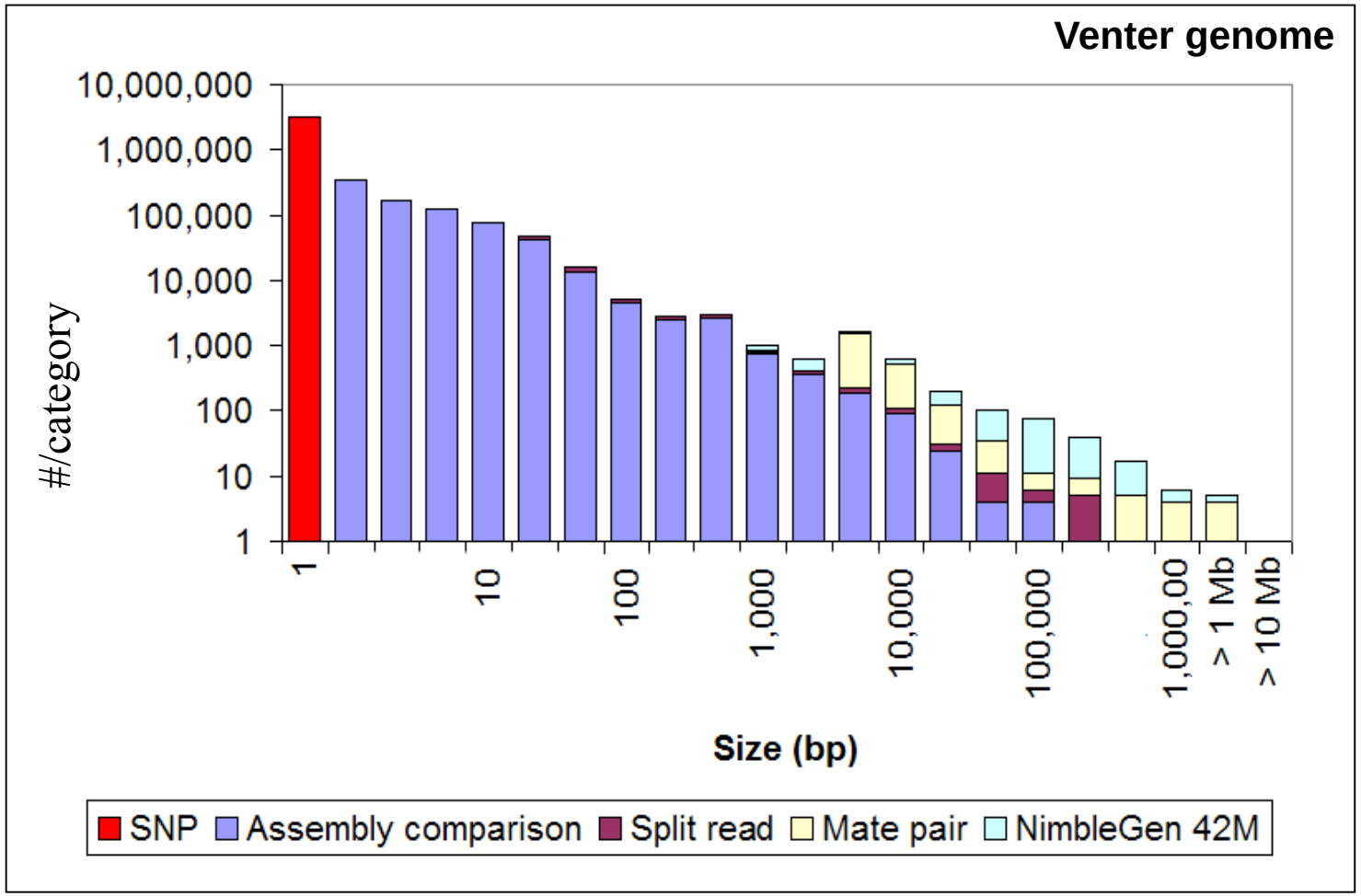
Gain





Unpublished data

How much genetic variation per genome?



-3,213,401 bp (0.11%) variable due to SNPs

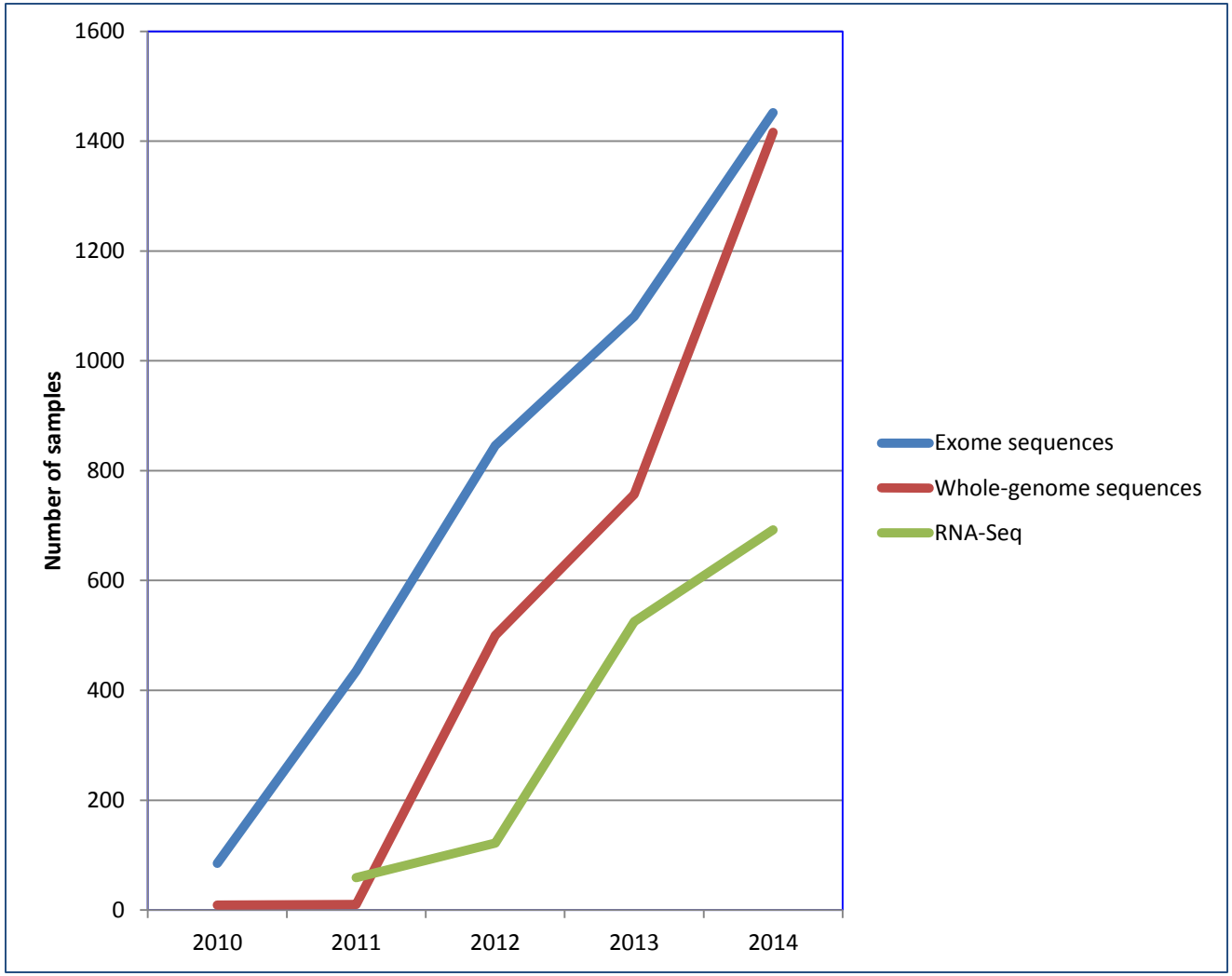
-40,568,593 bp (807,999 events; 1.35%) CNV/indel

-0.3% as inversion

-4,867 genes (~20%) impacted by CNV/indel

With Venter team
Science, 2003
Nature Genetics, 2006
PLOS Biology, 2007
Genome Biology, 2010
Human Mutation, 2013

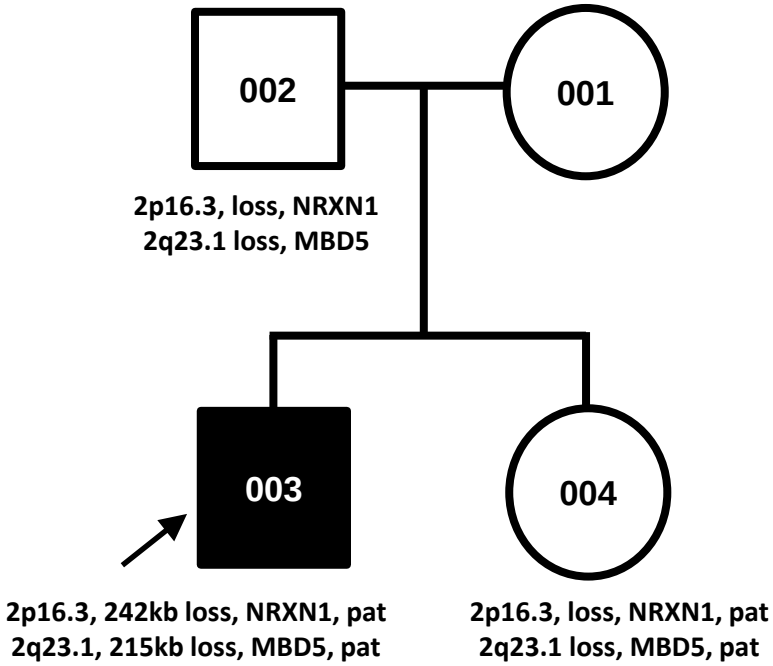
Clinical and Research: Shift to Genome-wide Analysis



Data from SickKids Hospital (DPLM + TCAG)

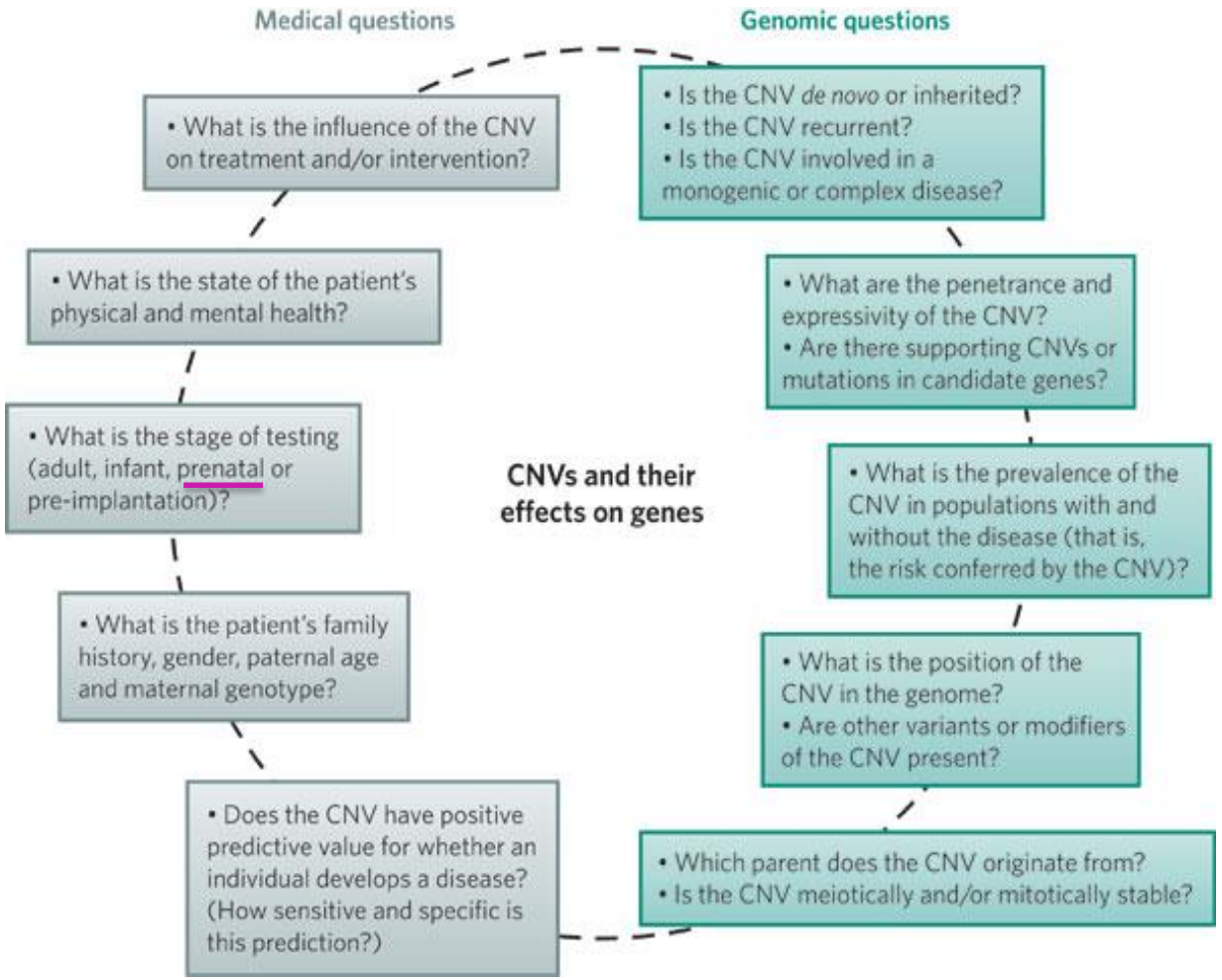
A Clinical Genomic Enigma Wrapped in a Question Mark?

(*especially in prenatal setting*)



…and a terrific research opportunity.

Clinical Context of CNVs
(and other genetic variants)



Nature, 2008