



Non-Coding Variation

**Fairmont Waterfront Hotel
Vancouver, BC, Canada**

18th October 2016

7:30 – 8:20 Registration

8:20 - 8:30 **Introduction**
Steven Brenner

Session 1 **Moderator: Steven Brenner**

8.30 – 9:00 **Functional Characterisation of Gene Regulatory Elements**
Nadav Ahituv
*Dept. of Bioengineering and Therapeutic Sciences, Univ.
California San Francisco, CA, USA*

9:00 – 9:30 **The Genomics of Human Physiological Adaptation**
Rasmus Nielsen
Univ. California Berkeley, CA, USA

9:30 – 9:45 **The Contribution of de novo Mutations in Enhancers and conserved Non-Coding Elements to Severe Developmental Disorders in 8000 Children**
Patrick Short
Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge CB10 1SA, UK.

9:45 - 10:00 **Tissue-Specific Functional Effect Prediction of Genetic Variation and Applications to Complex Trait Genetics**
Iuliana Ionita-Laza
Department of Biostatistics, Columbia University, New York, NY

10:00 – 10:15 Rapid Fire Poster Presentations

- Simon Fishilevich
- Marc Greenblatt
- Ping Liang
- Graham Taylor

10:15 – 11:00 Coffee Break & Poster Session

Session 2 Moderator: Bruce Gottlieb

11:00 – 11:30 Local and Distant Regulatory Effects of Genetic Variation in 44 Human Tissues in the GTEx Project
Barbara Engelhardt
Princeton Univ., NJ, USA

11:30 – 12:00 Genetic Variations in the Regulation of Alternative Splicing
Grace Xiao
Dept. Integrative Biology and Physiology, Univ. California Los Angeles, CA, USA

12:00 – 12:20 HGVS Nomenclature update
Reece Hart

12:20– 13:30 Lunch and HGVS Annual General Meeting

Session 3 BRCA Share Session
Moderator: Sean Mooney

13:30 – 13:40 Introduction
The Landscape of Organisations Dealing with BRCA

13:40 – 13:50 Origin of BRCA Share
Christophe Beroud
INSERM UMR_S910, Faculté de Médecine La Timone, 4ème étage, Marseille, France

13:50 – 14:15 Functional Assays as a Tool for Clinical Annotation of BRCA1 VUS
Nicholas Woods
Univ. California Berkeley, CA, USA

14:15 – 14:40 Massively Parallel Functional Analysis of Missense Mutations in BRCA1 for Interpreting Variants of Uncertain Significance
Lea Starita
Univ. Washington, WA, USA

Session 4 Moderator: Rachel Karchin

- 14:40 – 14:55** **Computational method to identify non-coding cancer drivers**
Ekta Khurana
Weill Cornell Medical College, New York, New York 10065, USA.
- 14:55 - 15:10** **Integrative analysis of epigenomes reveals roles of noncoding regions and variants in differentiation and diseases of blood cells**
Ross Hardison
Departments of Biochemistry and Molecular Biology and of Statistics, Pennsylvania State University, University Park, PA
- 15:10 – 15:40** **Title Not available at time of printing**
Olga Troyanskaya
Lewis Sigler Institute of Integrative Genomics, Princeton Univ., NJ, USA
- 15:40 – 15:45** Closing Remarks
- 15:45** **MEETING END IN TIME FOR ASHG PLENARY SESSION**