
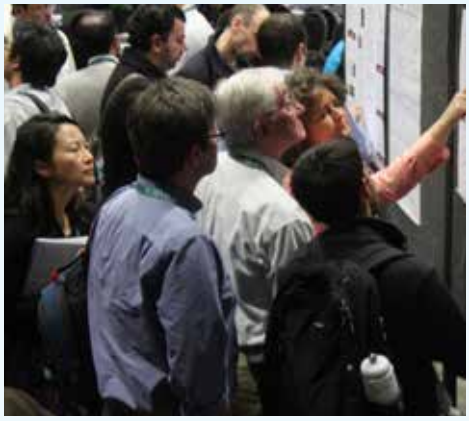


Join Keystone Symposia
for the 2016 conference on:

Understanding the Function of Human Genome Variation

May 31–June 4, 2016

Uppsala Konsert & Kongress | Uppsala | Sweden

Scientific Organizers:
Kerstin Lindblad-Toh and Xavier Estivill

One of the most complex problems in medical and evolutionary genomics is interpreting the function of the millions of variants the genome contains, most being rare and private to each individual or with consequences constrained to specific cells or tissues. The functional consequences of variation in coding regions are well established, but the majority of genetic variation resides in the noncoding portion of the human genome. The goal of this meeting is to bring together experts that may address important questions such as the function of noncoding variation, the connection between selection and disease, the diverse action of variants in different physiological and pathological scenarios, who develop and apply novel tools to connect genotype and phenotype both in disease and in an evolutionary context. By combining the diverse knowledge of many aspects of genomic analysis, we hope to bring out critical discussion and novel approaches to understanding human genome variation of crucial importance for the individualized genome analysis that precision medicine proposes. We are now entering the age of precision medicine with the capacity to analyze the genome of every subject, evaluating the functional consequences of variability and its interaction with the environment at different time-points in life.

Session Topics:

- Pleiotropy and Epistasis of Variants Involved in Disease
- Finding the Causative Variant(s)
- Connection between Selection and Disease
- Defining the Functional Elements in the Human Genome
- Human History, Migration and Evolution
- Selection and Population Genetics
- Complex Disease and Genetic Variation
- Structural Variation

Submitting an abstract is a great way of participating in the conference through poster presentation and possible selection for a short talk.

Scholarship & Discounted Abstract Deadline: Feb 1, 2016
Abstract Deadline: Mar 1, 2016
Discounted Registration Deadline: Mar 31, 2016

For additional details, visit **www.keystonesymposia.org/16K1**.

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TUESDAY, MAY 31

Arrival and Registration

WEDNESDAY, JUNE 1

Welcome and Keynote Address

Peter M. Visscher, University of Queensland, Australia
Estimation, Dissection and Understanding Human Genomic Variation for Complex Traits

Pleiotropy and Epistasis of Variants Involved in Disease

Len Pennacchio, Lawrence Berkeley National Lab, USA
Assessing Distant-Acting Enhancers in vivo

Michael Snyder, Stanford University School of Medicine, USA
Longitudinal Personal Genome Analysis

Jeanne Bentley Lawrence, University of Massachusetts, USA
Epigenetics and Genome Balance: Lessons from X-Chromosomes to Down Syndrome

Short Talk(s) Chosen from Abstracts

Finding the Causative Variant(s)

Xavier Estivill, Center for Genomic Regulation and University Pompeu Fabra, Spain
Convergence of Germ-Line Variants in Disease Susceptibility

Heidi Rehm, Harvard Medical School, USA
Resources to Support Variant Interpretation

Nuria Lopez-Bigas, University Pompeu Fabra - UPF, Spain
Defining Driver Mutations from Thousands of Cancers

Short Talk Chosen from Abstracts

Poster Session 1

THURSDAY, JUNE 2

Connection between Selection and Disease

Elinor Karlsson, University of Massachusetts Medical School, USA
Natural Selection and Cholera Resistance in Bangladesh

Joseph K. Pickrell, New York Genome Center, USA
Joint Analysis of Functional Genomic Data and Genome-Wide Association Studies of Human Traits

Kerstin Lindblad-Toh, Broad Institute of MIT & Harvard and Uppsala University
Exploring the Connection between Selection and Disease in Dogs and Humans

Barak Alon Cohen, Washington University School of Medicine, USA
Analysis of Combinatorial cis-regulation

Short Talk(s) Chosen from Abstracts

Defining the Functional Elements in the Human Genome

Gill Bejerano, Stanford University, USA
Finding 'Bugs' in the Human Genome Code

Ben Lehner, Centre for Genomic Regulation, Spain
The Effects of Genetic Variation in Space and Time

William J. Greenleaf, Stanford University, USA
ATAC-Seq - Chromatin Accessibility at the Single Cell Level

Short Talk Chosen from Abstracts

Poster Session 2

FRIDAY, JUNE 3

Human History, Migration and Evolution

Sarah A. Tishkoff, University of Pennsylvania, USA
Gene Conversion in Human Populations and the Disease Burden of Recessive Alleles

Svante Pääbo, Max Planck Institute for Evolutionary Anthropology, Germany
Functional Genomics of Ancient Hominids

Jada BennTorres, University of Notre Dame, USA
A Genetic History of Indigenous Caribbean Peoples

Ed Green, University of California, Santa Cruz, USA
The Footsteps of Human Prehistory Signals of Positive Selection

Short Talk(s) Chosen from Abstracts

Selection and Population Genetics

Tuuli Lappalainen, New York Genome Center & Columbia University, USA
Functional Variation of the Human Genome: Lessons from the Transcriptome

Cynthia M. Beall, Case Western Reserve University, USA
Selection for Adaptation to High Altitude in Human Populations

Carlos D. Bustamante, Stanford School of Medicine, USA
Impact of Ancestry and Admixture on Human Genomic Variation

Short Talk Chosen from Abstracts

Poster Session 3

SATURDAY, JUNE 4

Complex Disease and Genetic Variation

Cisca Wijmenga, University Medical Center Groningen, Netherlands
Mapping of Immune-Mediated Disease Genes

Michel A. J. Georges, University of Liège, Belgium
Cell Expression Profiles of Chronic Intestinal Inflammation

Iiris Hovatta, University of Helsinki, Finland
Modeling Genetic Variation and Anxiety in Mice

Nicole Soranzo, Wellcome Trust Sanger Institute, UK
Cardiovascular Disease and the UK10K Project

Short Talk(s) Chosen from Abstracts

Structural Variation

Evan E. Eichler, HHMI/University of Washington, USA
Evolution and Mechanisms of Gene Duplication and DNA Transposition within the Human Genome

Tomas Marques-Bonet, Universitat Pompeu Fabra/CSIC, Spain
Dynamics and Impact of Genomic Variation in Recent Human Evolution

Jonathan Sebat, University of California, San Diego, USA
CNVs in Neuropsychiatric Disease

Meeting Wrap-Up: Outcomes and Future Directions (Organizers)

* Session Chair † Invited but not yet accepted Program current as of ***October 21, 2015***. Program subject to change. Meal formats are based on meeting venue. For the most up-to-date details, visit ***www.keystonesymposia.org/16K1***.

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SUNDAY, JUNE 5

Departure