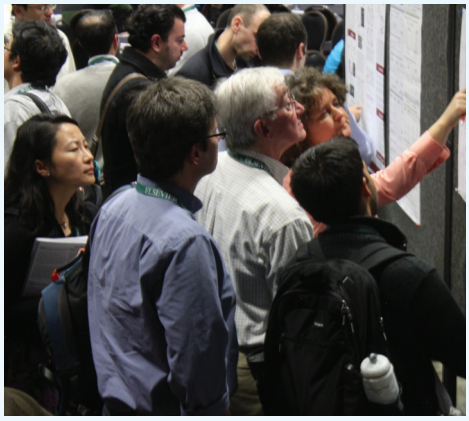

Join Keystone Symposia for the 2016 conference on:

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.

Abstract Deadline: Mar 15, 2016
Discounted Registration Deadline: Mar 31, 2016

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

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KEYSTONE SYMPOSIA
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Organized in collaboration with Knut and Alice Wallenberg Foundation and Science for Life Laboratory

Abstract & Scholarship Deadline: February 1, 2016 / Abstract Deadline: March 15, 2016 / Discounted Registration Deadline: March 31, 2016

TUESDAY, MAY 31

Arrival and Registration

WEDNESDAY, JUNE 1

Welcome and Keynote Address

*__Xavier Estivill__, Sidra Medical and Research Center, Qatar

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

*__Joseph K. Pickrell__, New York Genome Center, USA

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

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

Dana C. Crawford, Case Western Reserve University, USA
Phenome-Wide Association Studies

Alexandra E. Fish, Vanderbilt University, USA
Short Talk: Statistical Associations Suggest an Absence of Biological Epistasis in Human Gene Regulation

Laura Kasak, University of Tartu, Estonia
Short Talk: Somatic CNVs in the Placental Genome and Link to Pregnancy Success

Finding the Causative Variant(s)

*__Gill Bejerano__, Stanford University, USA

Xavier Estivill, Sidra Medical and Research Center, Qatar
Deconstructing Obsessive-Compulsive Disorder by Whole Exome Sequencing and Rare Variant Association Study

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

Nuria Lopez-Bigas, University Pompeu Fabra - UPF, Spain
Tumor Genomes Shed Light into Mutational Processes and Cancer Vulnerabilities

Fiona Allum, McGill University, Canada
Short Talk: Epigenome-Wide Association Studies of Blood Lipids Identify Tissue-Independent Disease Biomarkers

Seulkee Lee, KAIST, South Korea
Short Talk: Allele Imbalance Analysis Combined with Chromatin Interaction Data Predicts Functional Autoimmune Risk Loci

Poster Session 1

THURSDAY, JUNE 2

Connection between Selection and Disease

*__Tuuli Lappalainen__, New York Genome Center & Columbia University, USA

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

Joseph K. Pickrell, New York Genome Center, USA
Case-Control Association Mapping without Cases

Jessica Alfoldi, Broad Institute of MIT and Harvard, USA
Using Mammalian Conservation to Annotate Each Individual Base of the Human Genome – the 200 Mammals Project

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

Ryan N. Doan, Boston Children's Hospital, USA
Short Talk: Mutations in Human Accelerated Regions (HARs) are Associated with Abnormal Social and Cognitive Behavior

Kang Seon Lee, KAIST, South Korea
Short Talk: Evolutionary Acceleration in the Regulatory Sequences of Human Brain Development

Defining the Functional Elements in the Human Genome

*__Cynthia M. Beall__, Case Western Reserve University, USA

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

Daniel Zerbino, European Molecular Biology Laboratory, UK
Functional Annotation of Genomes at Ensembl

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

Kaur Alasoo, Wellcome Trust Sanger Institute, UK
Short Talk: Fine-Mapping Condition-Specific Regulatory Variants in Human Macrophages using ATAC-seq

Poster Session 2

FRIDAY, JUNE 3

Human History, Migration and Evolution

*__Tomas Marques-Bonet__, Universitat Pompeu Fabra/CSIC, Spain

Leif Andersson, Uppsala University, Sweden
The Genetics of Ruff Sex

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

Jada BennTorres, University of Notre Dame, USA
Genomic Perspectives on Indigenous Caribbean Histories in the Lesser Antilles

Mattias Jakobsson, Uppsala University, Sweden
The Genomic Footprints of Neolithic Europeans

Stephane Peyregne, Max Planck Institute for Evolutionary Anthropology, Germany
Short Talk: Detecting Ancient Positive Selection in the Human Genome from Extended Lineage Sorting

Martin Bodner, Medical University of Innsbruck, Austria
Short Talk: Complete Mitogenomes of Ancient Human Bones from the Northeastern Andes in South America

Selection and Population Genetics

*__Cisca Wijmenga__, University Medical Center Groningen, Netherlands

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

Cynthia M. Beall, Case Western Reserve University, USA
Hemoglobin Concentration and Reproductive Success of Tibetan Highlanders

Luis Quintana-Murci, Institut Pasteur, CNRS URA3012, France
Genetic and Evolutionary Determinants of Population Variation in Immune Responses

Corinne N. Simonti, Vanderbilt University, USA
Short Talk: High-Throughput Characterization of Phenotypic Effects of Recent Positive Selection and Archaic Introgression using Electronic Health Records Connected to Genotypes

Poster Session 3

SATURDAY, JUNE 4

Complex Disease and Genetic Variation

*__Peter M. Visscher__, University of Queensland, Australia

Cisca Wijmenga, University Medical Center Groningen, Netherlands
The Genetic Architecture of Cytokine Responses and their Association with Complex Diseases

Hyun Ji Noh, Broad Institute, USA
Short Talk: Shared Genetics of Compulsive Disorder in Dogs and Humans

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

Nicole Soranzo, Wellcome Trust Sanger Institute, UK
Genetic and Epigenetic Variation in Population Based Cohorts Informs Cardiometabolic and Immune Disease Risk

Ramy Arnaout, Beth Israel Deaconess Medical Center, Harvard Medical School, USA
Short Talk: Counting Missing Mutations: Estimating Cancer Diversity in the Overall Population from 10,000 Cancer Exomes

Linda Marie Boettger, Broad Institute, USA
Short Talk: Recurring Exon Deletions in the Haptoglobin (HP) Gene Associate with Lower Blood Cholesterol Levels

Structural Variation

*__Jessica Alfoldi__, Broad Institute of MIT and Harvard, USA

Evan E. Eichler, HHMI/University of Washington, USA
Duplications, Disease and the Evolution of the Human Genome

Tomas Marques-Bonet, Universitat Pompeu Fabra/CSIC, Spain
Upgrade on the Chimpanzee Reference Genome and Evolutionary Comparison of Tandem Repeat Variation

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

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

SUNDAY, JUNE 5

Departure