

# CHARGE

## (Cohorts for Heart and Aging Research in Genomic Epidemiology) Investigator Meeting Agenda

Los Angeles (Redondo Beach)  
Crowne Plaza Hotels & Resorts  
300 North Harbor Drive  
Redondo Beach, CA 90277  
January 21-25, 2014

### **Tuesday, January 21, 2014**

1:00pm-5:00pm Meeting Registration

### **Wednesday, January 22, 2014 – Day 1**

7:00am-1:30pm Meeting Registration and Continental Breakfast

#### **Session A: Individual Working Group Meetings**

8:00am-8:50am Blood Pressure working group – C. Liu/D. Levy  
Adiposity working group – I. Borecki  
Neurology/Stroke working group – S. Seshadri

8:50am-9:40am Hemostasis working group – N. Smith  
Gene Lifestyle working group – D.C. Rao  
Neurology/Dementia working group – S. Seshadri

*9:40am-10:00am Coffee Break*

10:00am-10:50am Gene Expression working group – M. Peter  
Subclinical working group – J. Bis  
Neurology/Cognition working group – S. Seshadri

10:50am-11:40am Commons working group – E. Boerwinkle  
Microbiome working group – R. Kraaij  
Hearing working group – P. Nagtegaal

*11:40am-1:10pm Lunch*

#### **Session B: Congress Session**

1:10pm-1:20pm Welcome  
Jerome I. Rotter (LABioMed/Harbor-UCLA)

1:20pm-1:35pm State of CHARGE  
Christopher O'Donnell (NHLBI)

<b>Session B1:</b>	<b>Cloud Computing and Genomics:</b> (Moderator – Lenore Launer)
1:40pm-2:00pm	Initial processing of CHARGE samples and using DNAnexus to support collaboration across CHARGE – Andrew Carroll (DNAnexus)
2:00pm-2:20pm	Cloud computing in CHARGE – Eric Boerwinkle (UT Houston)
<b>Session B2:</b>	<b>Working Group Presentations:</b> (Moderator – George Papanicolaou)
2:20pm-2:35pm	Neurology: Common Variants Associated with Intracranial and Hippocampal Volume – Hieab H.H. Adams (Erasmus MC, Rotterdam)
2:35pm-2:50pm	Mineral Metabolism: Common Genetic Variants Associated with Circulating Parathyroid Hormone Concentration – Cassianne Robinson-Cohen (University of Washington)
2:50pm-3:05pm	Chronic Kidney Disease: Novel Loci of Kidney Function: The CKDGen Consortium – Alexander Teumer (University Medicine Greifswald, Germany)
<i>3:05pm-3:35pm</i>	<i>Coffee Break</i>
<b>Session B3:</b>	<b>Tissues and Organelles:</b> (Moderator – Villmundur Gudnason)
3:35pm-3:55pm	Using GTEx to characterize genetic architecture and the genetic component to common diseases and complex humans – Nancy Cox (University of Chicago)
3:55pm-4:15pm	Analysis of mitochondrial DNA variants – Chunyu Liu (NHLBI)
<b>Session B4:</b>	<b>Family Studies:</b> (Moderator – Jerome I. Rotter)
4:15pm-4:35pm	A family-based search for high impact variants for glucometabolic traits – Donald Bowden (Wake Forest)
4:35pm-4:55pm	Linkage and association of EKG traits – Cornelia van Duijn (Erasmus MC, Rotterdam)
4:55pm-5:15pm	Inherited epigenetic effects on phenotype variation and disease risk – Joseph Nadeu (Pacific Northwest Diabetes Research Institute)
<b>Poster Session</b>	
5:30pm-7:00pm	Poster session (with wine and cheese)
<b>Evening</b>	
7:15pm	<i>RSC Dinner</i>
7:15pm	<i>Dinner on your own</i>

## **Thursday, January 23, 2014 – Day 2**

7:00am-1:30pm Registration and Continental Breakfast

### **Continue Congress Session**

**Session C1:** **Rare Variants:** (Moderators – Andre Uitterlinden and Adrienne Cupples)  
8:00am-8:20am Whole genome sequencing in CHARGE – Alanna Morrison (UT Houston)  
8:20am-8:40am Insights from whole-genome sequencing of 3,800 individuals and applications to disease – Brent Richards (McGill, Montreal)  
8:40am-9:00am Functional annotation of exome sequencing data – Jeroen van Rooij (Erasmus MC, Rotterdam)  
9:00am-9:20am Identification of the downstream consequences of disease-associated SNPs – Lude Franke (University Medical Center Groningen)

**Session C2:** **Current issues in rare variant analysis (or other issues), by the Analysis Committee Nominees:** (Moderators – Thomas Lumley and Joshua Bis)  
9:20am-9:40am Sharing individual participant data; how will it help – Kenneth Rice (University of Washington)  
9:40am-10:00am Rare variants analysis for binary traits in families – Qiong Yang (Boston University)  
10:00am-10:15am Meta-analysis of genome-wide association studies on fibrinogen levels using 1000G imputed data – Abbas Dehghan (Erasmus MC, Rotterdam)

10:15am-10:40am	Coffee Break
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**Session C3:** **Working group presentations:** (Moderator – Cashell Jaquish)  
10:40am-10:55am Glycemia–T2D: Exome Chip Analysis of 9,448 Type 2 Diabetes Cases and 48,728 controls Identifies Novel Rare and Low Frequency coding Variation in Known T2D Susceptibility Loci – Audrey Y. Chu (Brigham and Women’s Hospital, Boston)  
10:55am-11:10am Exome Chip: Loss-of-Function Mutations in the HAL Gene is Associated with Histidine Levels – Bing Yu (University of Texas, Houston)  
11:10am-11:25am Gene Expression: Direct Assessment of the Impact of Human Genes that are Differentially Expressed with Age on Longevity in *Caenorhabditis Elegans* – George L. Sutphin (The Jackson Laboratory, Bar Harbor, Maine)

**Session C3:** **Population vs Family Approaches:** (Moderator – Myriam Fornage)  
11:25am-11:45am The utility of families in understanding the genetic basis of type 1 diabetes – Stephen Rich (University of Virginia)

**Session C4:** **Rare Variants in Rare Disorders:** (Moderator – James Wilson)  
11:45am-12:05am Exome sequencing in rare disorders - Stanley Nelson (UCLA)  
12:05am-12:25pm Clinical applications of genomics – Wayne Grody (UCLA and ACMG)

**Session C5:** **CHARGE** (Moderator – Jerome I. Rotter)  
12:25pm-12:30pm CHARGE Infrastructure – Bruce Psaty (University of Washington)  
12:30pm-12:40pm CHARGE Business – Joshua Bis (University of Washington)  
12:40pm-12:50pm Group photo

*12:50pm-2:00pm Lunch*

**Session D:** **Individual Working Group Meetings**  
2:00pm-2:50pm CHARGE Analysis & CHARGE-S Analysis/Bioinformatics working group – K. Rice/A. Cupples  
Bone working group – D. Kiel  
Diabetes/Glycemia working group – J. Meigs  
2:50pm-3:40pm Genotyping/Exome chip/ENCODE working group – J. Rotter/C. O’Donnell  
Social Science working group – P. Koellinger  
Insulin-Like Growth Factor (IGF) working group – R. Kaplan

*3:40pm-4:00pm Coffee Break*

4:00pm-4:50pm Pharmacogenetics working group – B. Psaty  
Lipids working group – A. Cupples  
Pulmonary working group – S. London  
4:50pm-5:40pm Family Studies working group – D. Siscovick/C. O’Donnell  
Nutrition working group – C. Smith  
Epigenetics working group – M. Fornage/L. Stolk  
5:40pm-6:30pm Inflammation working group – B. Alizadeh

*5:40pm-6:30pm Break*

**Evening**  
6:30pm Evening Event (Entire congress), at the Crowne Plaza

**Friday, January 24, 2014 – Day 3**

**Joint ENCODE/CHARGE Meeting**

**Session D (continued): Individual Working Group Meetings**

7:00am-7:50am Neurology/MRI – S. Seshadri

7:00am-11:30am Registration and Continental Breakfast

**Session E1: ENCODE**

8:00am-8:05am Introduction to Workshop – David Siscovick (University of Washington)

8:05am-8:20am Introduction to ENCODE – Elise Feingold (NHGRI)

**Opening talks** (Moderator – Elise Feingold, NHGRI)

8:20am-8:50am Introduction to ENCODE and future perspective – Michael Snyder (Stanford University)

8:50am-9:20am Interpreting ENCODE RNA data – Barbara Wold (Caltech)

9:20am-9:50am Interpreting ENCODE chromatin/regulation data – John Stamatoyannopoulos (University of Washington)

9:50am-10:20am Strategies for integrating ENCODE and GWAS data – Robert Klein (Memorial Sloan-Kettering Cancer Center)

*10:20am-10:45am Coffee Break*

**Session E2: ENCODE and CHARGE**

10:45am-11:15am Regulatory variants associated with maternal metabolism – Timothy Reddy (Duke University)

11:15am-12:00noon Panel Q&A (Moderator – Jerome I. Rotter)

ENCODE: Mike Snyder, Barbara Wold, John Stamatoyannopoulos, Robert Klein, Timothy Reddy

CHARGE: Eric Boerwinkle, Jennifer Brody, Cornelia Van Duijn, Josée Dupuis

*12:00pm-1:15pm Lunch*

**Session F1: ENCODE/CHARGE Workshop (Group Presentation)**

1:15pm-1:20pm Introduction to the afternoon session – Robert Klein

1:20pm-1:50pm

Introduction to metadata – Eurie Hong and Cricket Sloan (Stanford University)

*In this session, an overview of the types of experimental details that are curated by the labs and the Data Coordinating Center (DCC) will be presented, including a general description of why we do it – why it's important for data consistency, searching, and future integrative studies. This will further include an overview of the [encodeproject.org](http://encodeproject.org) website, UCSC Genome Browser, ENSEMBL, Roadmap Epigenomics websites, etc. about how you can search for data using the metadata including discussion of what the actual cell lines available are.*

## **Session F2: ENCODE/CHARGE Workshop (Individual Tables)**

*Different members of the ENCODE consortium will be at different tables around the room focusing on various topics of interests. These tables are designed to facilitate informal discussions about how to access and use the ENCODE data; participants are urged to circulate among the tables.*

*Tables marked with a \* are designed for people interested in working with the data on a text file level using their favorite programming tools; all other tables are geared towards those most interested in navigating the ENCODE data and conducting analyses in their web browser. Topics include:*

- **Metadata**

1. Introduction to browsing the metadata - Eurie Hong (Stanford University). This table will enable hands-on exploration of the metadata that describes what is available in ENCODE through the web browser.
2. Introduction to computing on the metadata\* - Cricket Sloan (Stanford University). This table will enable hands-on exploration of the metadata that describes what is available in ENCODE through the web browser through downloading and parsing the underlying files.

- **Introduction to ENCODE data types.**

*These sessions will include illustrations of how to access the various types of data from ENCODE (RNAseq, ChIP-Seq, GENCODE, etc.) and the various parameters and options for each. These will include demos of the new DCC website for searching these data.*

3. RNASeq and GENCODE – Leonard Lipovich (Wayne State University)
4. Regulatory elements including ChIP-Seq and DNase-Seq data – Anshul Kundaje (Stanford University)
5. RNA-Seq\* - Joel Rozowsky (Yale University)
6. DNase-Seq and ChIP-Seq, including use of BEDOPS\* - Matt Maurano (University of Washington)

- **Interpreting GWAS hits with ENCODE data.**

*This will be an overview of integrative analysis of ENCODE data that can be searched to annotate variants/GWAS*

7. HaploReg, RegulomeDB, and ENSEMBL VEP – Luke Ward (MIT)
8. Use of BEDOPS/BEDTOOLS specifically with GWAS data as well as other issues in analyzing GWAS data with ENCODE\* - Gosia Trynka (Brigham and Women's Hospital) and James Hayes (Memorial Sloan-Kettering Cancer Center)

1:55pm-2:35pm            Workshop Table Session 1

2:35pm-3:15pm            Workshop Table Session 2

3:15pm-3:35pm	<i>Break</i>
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3:35pm-4:15pm            Workshop Table Session 3

4:15pm – 4:45pm            General conversations focused at the tables to develop and catalyze potential collaborations. This time is especially well suited for CHARGE investigators with specific scientific questions about their GWAS results to interact with the specific ENCODE investigators who have the expertise to aid in the analysis of the regions, thereby sparking future collaborations.

4:45pm-5:00pm            Wrap-up: CHARGE working with ENCODE, ENCODE working with CHARGE – David Siscovick and Robert Klein

**Evening**

5:30pm-7:00pm            *Closing reception (wine and cheese)*

7:00pm                        *Dinner on your own*

**Saturday, January 25, 2013**

Depart