

# Human Genetic Variation and Disease

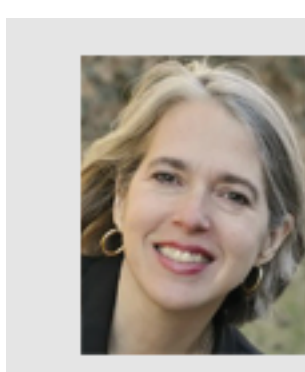
## Scientific program

### Sunday

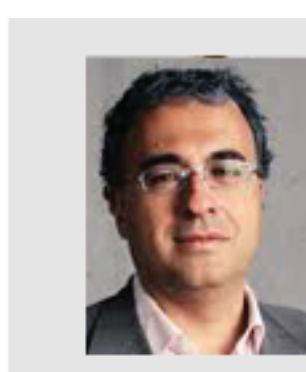
### WELCOME

#### Session 1 Keynote

#### What is the future of human variation and disease science?



**Discussion Leader:**  
Elizabeth Phimster,  
Deputy Editor,  
New England Journal  
of Medicine,  
Boston, MA, USA



**Speaker:**  
Roderic Guigo,  
Universitat Pompeu  
Fabra, Barcelona,  
Spain

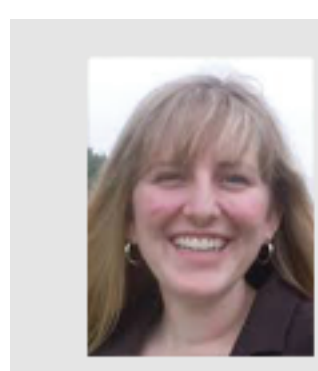


**Speaker:**  
Evan Eichler,  
University of Washington,  
Seattle, USA

### Monday

#### Session 2

#### Emerging classes of variation and disease



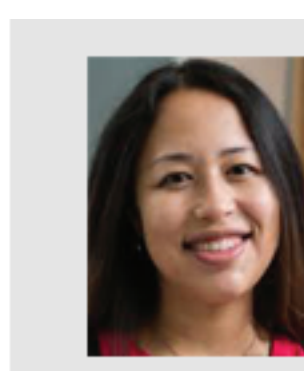
**Discussion Leader:**  
Tychele Turner,  
University of  
Washington,  
Seattle, USA



**Speaker:**  
Kai Tan,  
Children's Hospital  
of Philadelphia, USA



**Speaker:**  
Ekta Khurana,  
Weill Cornell Medical  
College,  
New York City, USA



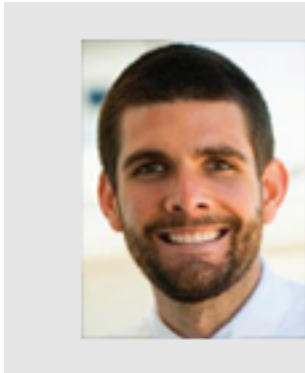
**Speaker:**  
Angela Brooks,  
University of California  
Santa Cruz, USA



**Speaker:**  
Rob Scharpf,  
Johns Hopkins  
University Medicine,  
Baltimore, USA

#### Session 3

#### Genetic variation in cancer



**Discussion Leader:**  
Mark Saoum,  
Personal Genome  
Diagnostics,  
Baltimore, USA



**Speaker:**  
Hannah Carter,  
University of California  
San Diego, USA

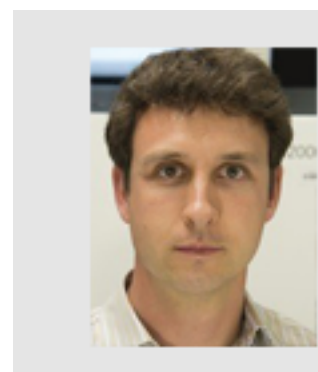


**Speaker:**  
Alfonso Valencia,  
Barcelona  
Supercomputing Center,  
Spain

### Tuesday

#### Session 4

#### Data-driven approaches for precision medicine



**Discussion Leader:**  
Obi Griffith,  
Washington University,  
St. Louis, USA



**Speaker:**  
Wolfgang Huber,  
European Molecular  
Biology Laboratory,  
Heidelberg, Germany



**Speaker:**  
Michael Beer,  
Johns Hopkins  
University Medicine,  
Baltimore, USA



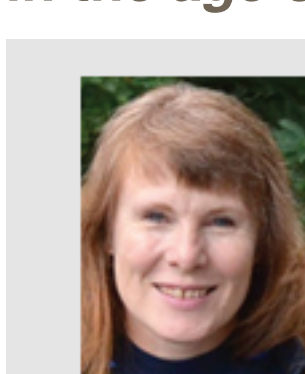
**Speaker:**  
Ben Raphael,  
Princeton University,  
USA



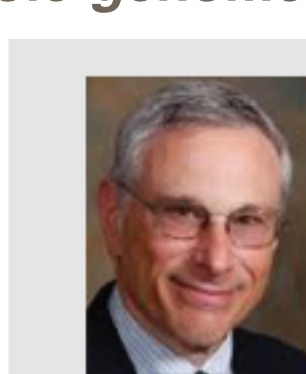
**Speaker:**  
Luis Serrano,  
Center of Regulatory  
Genomics,  
Barcelona, Spain

#### Session 5

#### Genetic testing in the age of whole-genome and whole-exome sequencing



**Discussion Leader:**  
Melissa Cline,  
University of California  
Santa Cruz, USA



**Speaker:**  
Robert Nussbaum,  
Invitae, San Francisco,  
USA

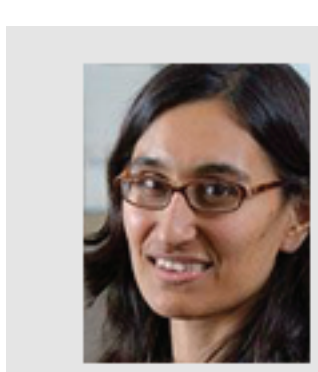


**Speaker:**  
Heidi Rehm,  
Harvard Medical School,  
Boston, USA

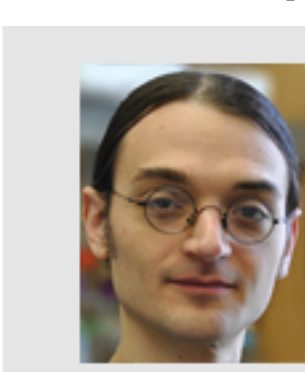
### Wednesday

#### Session 6

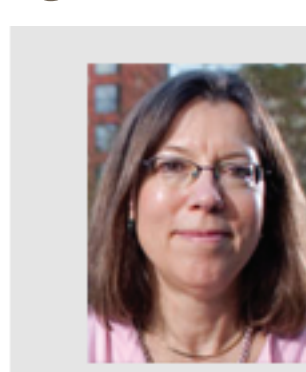
#### New directions: ribosome profiling, microbiomes, single cell technologies



**Discussion Leader:**  
Christina Leslie,  
Memorial Sloan  
Kettering Cancer  
Center, New York City,  
USA



**Speaker:**  
Nicholas Ingolia,  
University of California  
Berkeley, USA



**Speaker:**  
Cynthia Sears,  
Johns Hopkins  
University Medicine,  
Baltimore, USA



**Speaker:**  
Andrew Clark,  
Cornell University,  
Ithaca, USA



**Speaker:**  
Smita Krishnaswamy,  
Yale University,  
New Haven, USA

#### Session 7

#### Common variation and disease



**Discussion Leader:**  
Michael Hicks,  
Human Longevity,  
San Diego, USA



**Speaker:**  
Marcella Devoto,  
Baylor College of  
Philadelphia, USA



**Speaker:**  
Francesco Cucca,  
Cittadella Universitaria  
di Monserrato, Italy

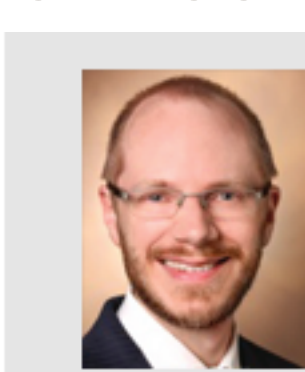
### Thursday

#### Session 8

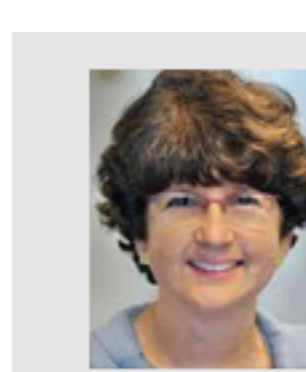
#### Integrating genomic variation with medical records



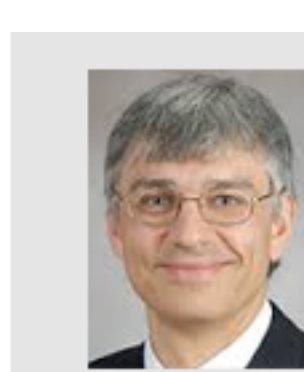
**Discussion Leader:**  
Brian Shirts,  
University of Washington,  
Seattle, USA



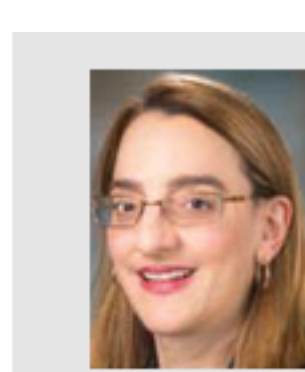
**Speaker:**  
Joshua Denny,  
Vanderbilt University,  
Nashville, USA



**Speaker:**  
Sharon Plon,  
Baylor College of  
Medicine, Houston, USA



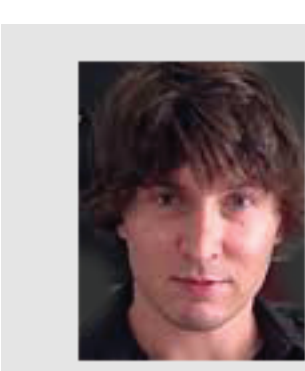
**Speaker:**  
Elmer Bernstam,  
University of Texas  
Health Science Center,  
Houston, USA



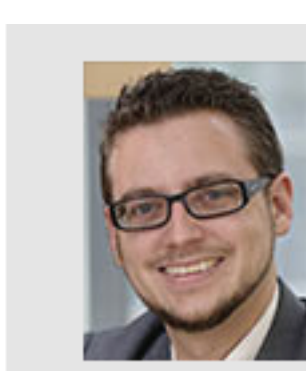
**Speaker:**  
Funda Meric-Bernstam,  
M.D. Anderson,  
Houston, USA

#### Session 9

#### Biophysical perspectives on variation and disease



**Discussion Leader:**  
David Masica,  
Johns Hopkins  
University,  
Baltimore, USA



**Speaker:**  
John Chodera,  
Memorial Sloan  
Kettering Cancer Center,  
New York City, USA



**Speaker:**  
Anna Pachenko,  
National Center for  
Biotechnology  
Information,  
Bethesda, USA

### Sat / Sun

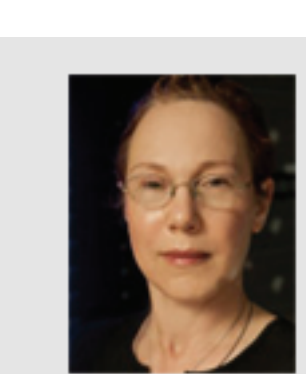
### GRS Gordon Research Seminar

By and for young scientists -  
opportunities and challenges inherited by  
the next generation of researchers.

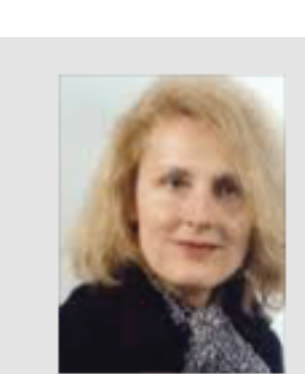
### Monday

### POWER HOUR

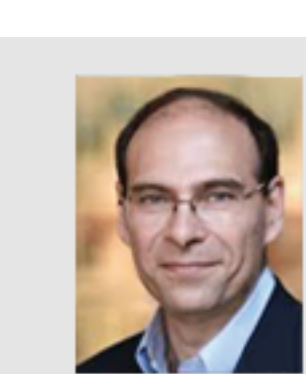
Open discussion to support the professional  
growth of URMs and women in our community  
With Rachel Karchin & Anna Pachenko



**Chair:**  
Rachel Karchin,  
Johns Hopkins  
University,  
Baltimore, USA



**Chair:**  
Rita Casadio,  
University of Bologna,  
Italy



**Vice Chair:**  
Olivier Lichtarge,  
Baylor College of  
Medicine,  
Houston, USA



**Vice Chair:**  
Yana Bromberg,  
Rutgers University,  
Newark, USA



**GRS Chair:**  
Vikas Pejaver,  
University of Washington  
Seattle, USA