Large Seminar Room 8th Floor

3:30	**Parametric Human Project** Azam Khan Autodesk We introduce the Parametric Human Project, a not-for-profit organization building an eScience platform for multiscale digital human modeling. By encoding and centralizing research observations and results into a common data architecture, we hope to enable multiscale systems modeling from whole organism to organic molecule.
3:45	**Quantification of Genomic Uniqueness** Ajay Royyuru IBM A potential concern of research subjects in genomic studies is the risk of reidentification and resulting loss of genetic privacy. We describe an approach to quantification of uniqueness of given genomic data, taking into account the statistics of incidence across populations. Such measure of genomic uniqueness could be included in the informed consent process. We also suggest the development of an open standard (data and tools) to implement this widely.
4:00	**Dissecting human diploidy: Results from up to 1092 genomes.** Margret Hoehe Max Planck Institute for Molecular Genetics, Berlin Human genomes are diploid: have two sets of chromosomes, one from the mother, one from the father, and therefore encode two forms of each gene, protein and non-coding functional sequence. Key questions are: Which ones of these homologues are the same, which ones are different? How are the genetic variants distributed between the two chromosomal homologues?
4:15	**Democratizing Rare Cancer Genomics Research Using Social Media** Corrie Painter Broad Institute Rare cancers are often neglected by the biomedical community do to lack of funding, and lack of centralized tissue. The era of social media has restructured the way that patients interact, and has provided a platform upon which patients with rare cancers can form organic communities. As proof of

	concept that directly engaging patients though social media can democratize cancer genomics research, the Broad Institute is launching a direct to patients project in an exceedingly rare cancer, angiosarcoma, where we will perform tumor/normal WES and RNAseq and couple the data with clinically annotated patient samples.
4:30	**COMPARE: COllaborative Management Platform for detection and Analyses of (Re-) emerging and foodborne outbreaks in Europe** Esther Pronker RIVM.NL Overview of the COMPARE initiative and more details on Work Package 12: investigating the barriers for open-source sharing of real-time/rapid WGS/metadata for outbreak detection and analysis Political, ethical, legal, administrative and regulatory barriers to sharing whole genome sequences, focussing on pathogen genetic sequences and human metadata
4:45	**Repositive - a free online tool for finding human genomic data sets from repositories and institutions around the world** Fiona Nielsen Find the datasets that you have been searching for in places that you did not know and comment on data that you have made available. The Repositive platform is now available in beta testing - sign up via http://repositive.io
5:00	**The capabilities of Population Genome Graphs, a novel approach to the concept of reference genomes** Sebastian Wernicke - Managing Director UK of Seven Bridges Genomics It would be exciting to discuss the capabilities of Population Genome Graphs, a novel approach to the concept of reference genomes that can incorporate a wide range of genetic variations and population diversity into a unified, compact representation. When used as a reference genome, Population Genome Graphs hold the promise to significantly increase the rate and quality of variant identification from sequence data. Used as an integrated view on a collection of genomes, they promise to vastly improve capabilities for the interpretation of variant data as well as its collection, storage, and dissemination. While these advantages over the current paradigm are clear in theory, it has been a topic of debate whether Population Genome Graphs can be efficiently implemented in

	practice. With significant support from Genomics England, we have built a full pipeline to build and process Population Genome Graphs and are happy to share real-world experimental results and considerations in advance of their publication.
5:15	**openSNP** Bastian Greshake our lessons of four years of doing open source personal genomics In 2011 we started accepting personal genomics data donations with openSNP. Now, four years and over 2000 data sets later, it's time to reflect on some lessons learned.
5:30	**Measuring the Outcomes of Predispositional Genome Sequencing in Healthy Adults: The PeopleSeq Study and Collaboration** Michael Linderman Multiple clinical, research and commercial programs are returning reports and or genomic data from personal genome sequencing (PGS) to ostensibly healthy adult participants. Only a little is known yet about the short and long-term outcomes of PGS in this context. The PeopleSeq project is a collaborative longitudinal study of PGS outcomes for participants in multiple cohorts, including PGP Harvard. In this talk I will describe the study goals and briefly present the results collected to date.
5:45	**Global Network of Personal Genome Projects -- UPDATES** Harvard PGP PGP Canada PGP-UK Genom Austria Resilience Project @ Mt Sinai

.