

BIG seminar 2011-2012

Biology and integrative genomics

**Monday,
7 November
17h**



Evan Eichler
University of Washington

”Structural variation, disease and evolution of the human genome.”

I will discuss the evolution of duplicated sequences in the human genome and how these genomic hotspots allowed us to discover pathogenic copy number variation associated with autism, intellectual disability and epilepsy. The study of this variation suggests that rare variation compounds to affect phenotypic outcome. Using next-generation sequence datasets, we can now quantify genetic variation in these duplicated genes allowing us to assess their contribution to disease and evolution.