**Background**

CNVs and methods for testing CNV association

Bayesian linear regression with sparse prior

Understanding the proposed model

**Data collection and explorative analysis**

Schizophrenia CNV data: both de novo and case-control [Pocklingtong, Neuron, 2015], [Szatkiewicz, Mol Psychiatry, 2014]

Autism CNV data: [Pinto, AJHG, 2014], [Sanders, Neuron, 2016]

Developmental delay CNV data (downloaded by Nick) [Coe, NG, 2015]

Segmentation of disjoint CNV regions

Pattern of overlapping CNVs

Burden analysis of CNVs

**Simple model for independent CNVs**: both de novo & case-control.

Creating independent CNVs: focus on most significant CNVs, or merge highly similar CNVs

Model of independent de novo CNVs: gene-level evidence.

Model of independent case-control CNVs.

Simulation: assessing validity of BFs.

Evaluation.

**Model development for overlapping inherited CNVs**

Model.

Simulation.

Application in SCZ case-control CNV data

Evaluation

## Reference

Human copy number variation and complex genetic disease [Girirajan & Eichler, ARG, 2011]

Functional impact of global rare copy number variation in autism spectrum disorders. [Pinto & Scherer, Nature, 2010]

Accurately Assessing the Risk of Schizophrenia Conferred by Rare Copy-Number Variation Affecting Genes with Brain Function [Raychaudhuri & Daly, PLG, 2010] // methodology paper

Convergence of genes and cellular pathways dysregulated in autism spectrum disorders [Pinto & Scherer, AJHG, 2014]

Refining analyses of copy number variation identifies specific genes associated with developmental delay [Coe & Eichler, NG, 2014]

Copy number variation in schizophrenia in Sweden [Szatkiewicz, Mol Psychiatry, 2014]

Novel Findings from CNVs Implicate Inhibitory and Excitatory Signaling Complexes in Schizophrenia, [Pocklingtong, Neuron, 2015] // methodology paper

A New Method for Detecting Associations with Rare Copy-Number Variants [Tzeng, PLoS Genetics, 2015]

Insights into autism spectrum disorder genomic architecture and biology from 71 risk loci [Sanders, Neuron, 2016]