

Case studies



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Workshop outline

- Introduction
- Omic data – genetic, epigenetic and metabolite
- Prediction – methodology
- Prediction – demonstration
- **Examples and case studies**



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Outline

- In this section we're going to cover examples of prediction using 'omics datasets including:
 - Genetics
 - Metabolomics
 - Epigenetics
 - Multi-omics

Prediction using genetics

- Polygenic scores: The sum of “risk” alleles for a particular trait
 1. GWAS in sample 1
 2. Genotype data in sample 2
 3. Weight the alleles in sample 2 by the beta coefficient from sample 1 (optional)
 4. Add up all the "risk" alleles in sample 2
- Often variants selected are "independent" and by a P value threshold cutoff



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Prediction using genetics

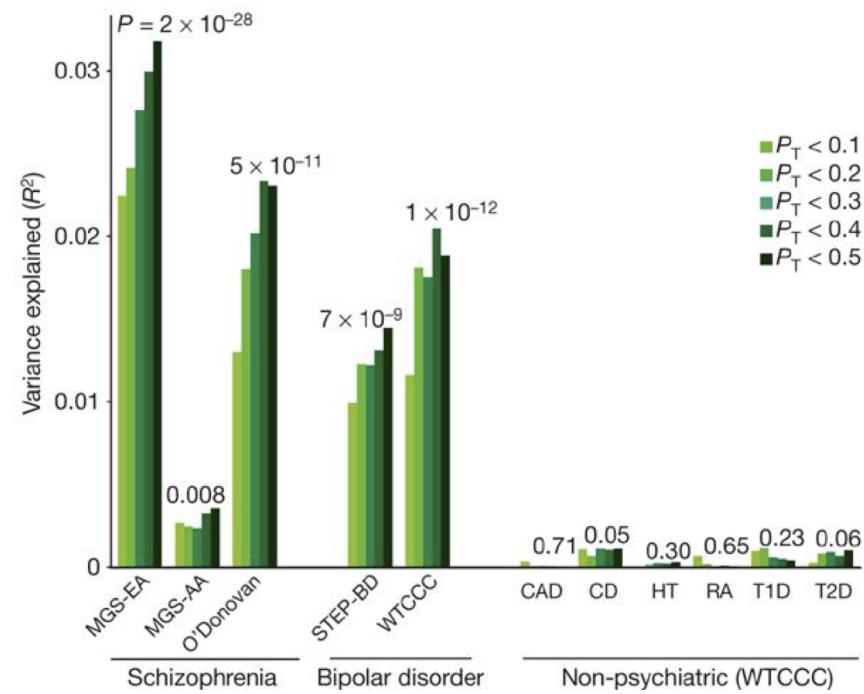
- Use of polygenic scores or genetic scores
- Simple prediction e.g. linear/logistic regression and AUC/C-statistics
- Often poor AUCs... (<0.7)

First one in 2009!

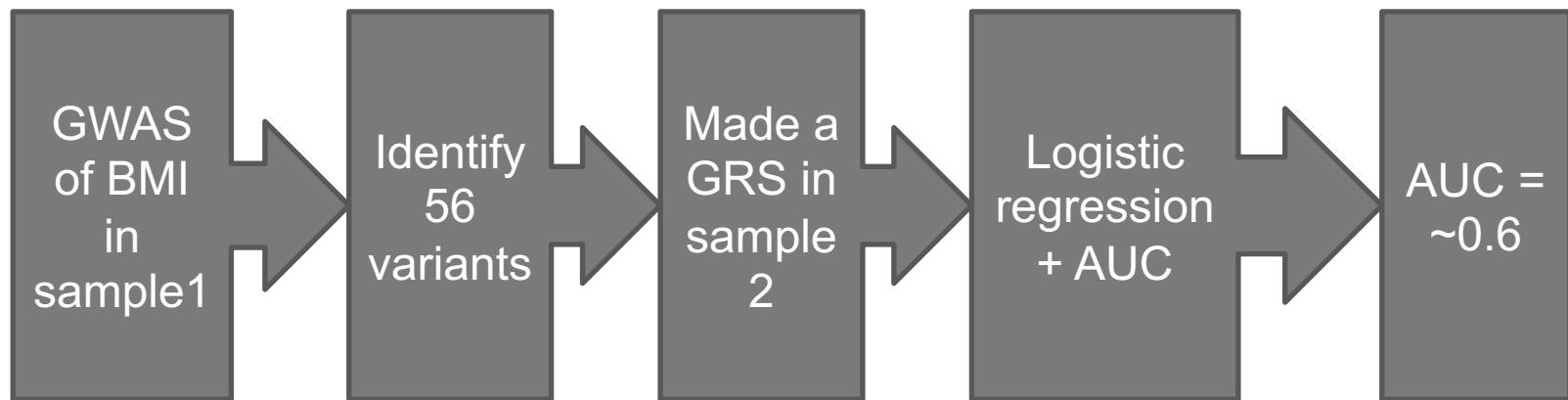
Common polygenic variation contributes
to risk of schizophrenia and bipolar
disorder

The International Schizophrenia Consortium

Nature 460, 748–752 (06 August 2009) | Download Citation ↴



Genetic risk sum score comprised of common polygenic variation is associated with body mass index



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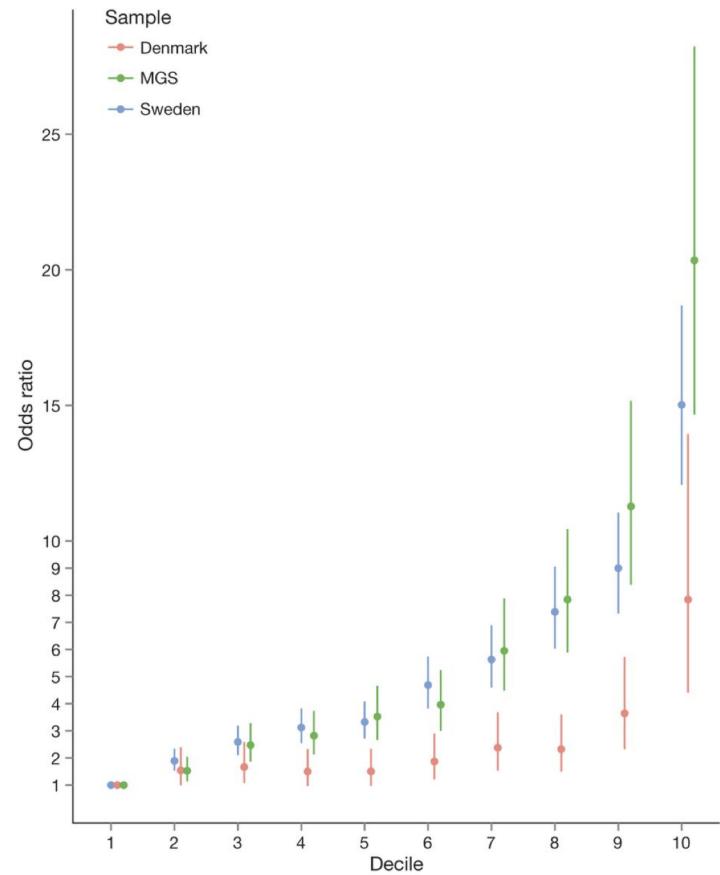


Use of genetic variants as a predictor

Biological insights from 108 schizophrenia-associated genetic loci

Schizophrenia Working Group of the Psychiatric Genomics Consortium

“Nevertheless, we stress that the sensitivity and specificity of RPS do not support its use as a predictive test. For example, in the Danish epidemiological sample, the area under the receiver operating curve is only 0.62”



Conceptual difference

- Germline variants are fixed at conception!

