

DOCTORAL SCHOOL BIOMEDICAL SCIENCES

## (Pharmaco)genetics in schizophrenia

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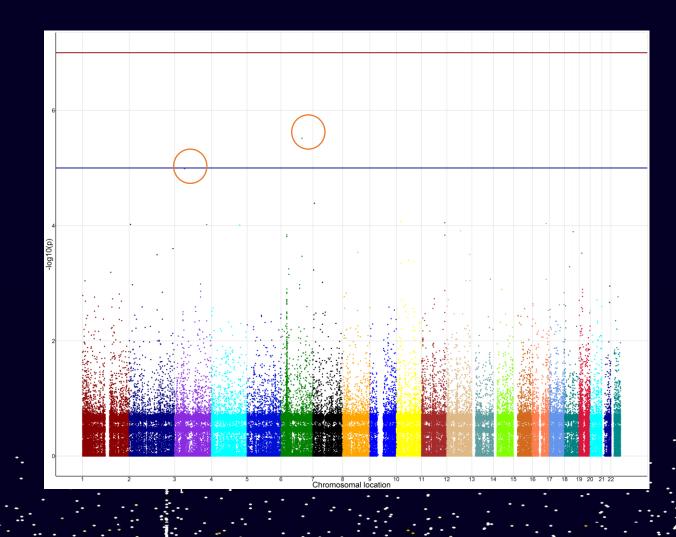




## Case-control study of exonic variation

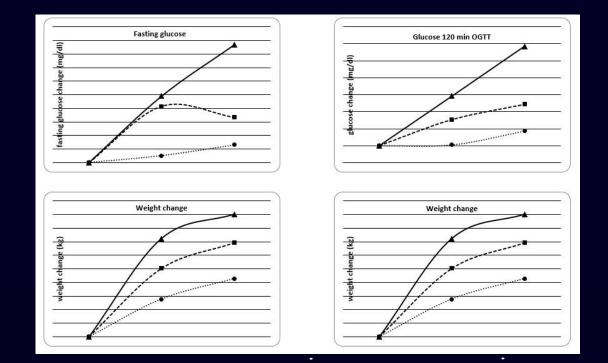
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- 493 patients with schizophrenia or schizoaffective D
- 484 healthy controls
- No significant findings
  - Best 2 genes
    - WISP3 (6q21 region):
      rs1230345 (p=3.05\*10<sup>-6</sup>)
    - CACNA2D3
      - rs9311525 (p=1.03\*10<sup>-6</sup>) and rs1558557 (p=3.85\*10<sup>-5</sup>)



## Genetic influence on metabolic syndrome

- Genetic association studies:
  - MTHFR
    - Prospective design
    - C allele of A1298C had the worst metabolic outcomes
  - Circadian clock and involvement in metabolic syndrome
    - Cross-sectional study
    - NR3C1 rs6196 A allele carriers had worse metabolic parameters



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## IGF variation and DNA methylation

- Cross-sectional study of genetic and epigenetic variantion
  - DNA variation:
    - 27 SNPs in Insulin Growth Factors
    - 7 differentially methylated regions (Methylation as epigenetic marker)
  - No significant findings related to metabolic outcomes
  - But: clear association between genetic variation and methylation
- Note: Current literature on (hypothesis driven) genetic association studies