Introduction to
Penn Neurodegeneration Genomics Center
(PNGC)

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Scientific Mission

Use genetics from GWAS and next generation sequencing studies to understand neurodegenerative diseases and identify therapeutic targets.
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<th>Areas of research</th>
<th>Details</th>
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<td>Gene discovery</td>
<td>- GWAS, linkage, and DNA sequencing</td>
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<td>Analysis</td>
<td>- Methodologies for complex disease genetics</td>
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<td>Bioinformatics</td>
<td>- Bioinformatics approaches to complex disease</td>
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<td>Functional genomics</td>
<td>- Translate loci discovery to gene discovery, and learn disease gene regulation</td>
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<td>Systems biology</td>
<td>- Network-based approaches for genetic findings</td>
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<td>Model organisms</td>
<td>- Screen candidate loci and functional analysis</td>
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| Risk prediction / endophenotypes | - Deep phenotype data from multiple cohorts  
                                 | - Endophenotype analysis and risk prediction |
PNGC is the national hub of AD genetics

- **ADGC (Cohort expansion)**
  - AD Genetics Consortium
  - >35,000 subjects
  - Found ~20 new AD genes

- **CASA (Sequencing analysis)**
  - Consortium for AD Sequence Analysis
  - Largest ADSP analysis team

- **CGAD (National coordination)**
  - Center for Genetics/Genomics of AD
  - Analyze data nationwide

- **NIAGADS (Data Resource)**
  - NIA genetics of AD data storage site
  - National repository for AD genetics data

- **PNGC programs form the national hub for AD genetics and drive the Alzheimer’s Disease Sequencing Project (ADSP), a White House/NIH initiative**
  - NIH committed ~$70M to these projects to build new cohorts, coordinate analysis, and disseminate data
Resources

- Computing capacity
  - Genomics database
  - Expertise and capacity for Amazon Cloud
  - 2PB storage (tape/hd)

- AD genomics data
  - AD GWAS (n=36,000)
  - WES (n=12,000)
  - WGS (n=4,000; 2017)
Center members

• Penn faculty with research interests relevant to the scientific mission of the center are welcome to join
• Members attend seminars/meetings, collaborate on research, contribute to internal core program, participate in training
• Currently 12 members/6 departments
  – Pathology (Brad Johnson / Virginia Lee / Gerard Schellenberg / John Trojanowski / Li-San Wang)
  – Biostat & Epidemiology (Mingyao Li / Adam Naj)
  – Genetics (Casey Brown / Chris Stoeckert)
  – Statistics (Nancy Zhang)
  – Pharmacology (Ben Voight)
  – Radiology (Christos Davatzikos)
• Website: www.med.upenn.edu/pngc (soon to be released)