The NINDS Repository Biomarker Discovery Collection: A Public Resource of Biomaterials for Neurodegenerative Disease Research

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The Coriell Institute

• History
  – Founded in 1953, Camden, NJ
  – Independent, not-for-profit organization

• Biobanking
  – House major NIH-funded and private biorepositories
  – > 2 million vials of cells
  – > 2 million tubes of DNA

• Distribution
  – 8,000 cell cultures, 100,000 DNA specimens each year
  – 64 countries
  – > 7,000 peer-reviewed papers citing ~ 12,000 biospecimens
The NINDS Repository

- Founded in 2002 with the goal of accelerating genetic discoveries in complex neurologic disorders
- Includes biological samples from > 41,000 individuals
- Biospecimens and clinical data available via online catalog ([http://ccr.coriell.org/NINDS/](http://ccr.coriell.org/NINDS/))

<table>
<thead>
<tr>
<th>Disease Subcollection</th>
<th>Samples Received</th>
<th>Unique Subjects</th>
<th>Affected Subjects</th>
<th>At Risk Subjects</th>
<th>Unaffected Subjects</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alzheimer’s Disease</td>
<td>6</td>
<td>4</td>
<td>2</td>
<td>2</td>
<td>0</td>
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<tr>
<td>Cerebrovascular Disease</td>
<td>20,422</td>
<td>12,478</td>
<td>10,285</td>
<td>2,193</td>
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<tr>
<td>Control</td>
<td>13,892</td>
<td>6,958</td>
<td>0</td>
<td>0</td>
<td>6,958</td>
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<tr>
<td>Dystonia</td>
<td>1,862</td>
<td>926</td>
<td>910</td>
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<tr>
<td>Epilepsy</td>
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<td>5,650</td>
<td>3,819</td>
<td>1,831</td>
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<tr>
<td>Huntington's Disease</td>
<td>25,406</td>
<td>1,428</td>
<td>256</td>
<td>860</td>
<td>412</td>
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<tr>
<td>Motor Neuron Disease</td>
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<td>2,743</td>
<td>2,539</td>
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<tr>
<td>Parkinsonism</td>
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<td>6,135</td>
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<td>Tourette Syndrome</td>
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<td>4,051</td>
<td>2,846</td>
<td>1,205</td>
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<tr>
<td><strong>TOTAL</strong></td>
<td><strong>100,081</strong></td>
<td><strong>41,123</strong></td>
<td><strong>26,792</strong></td>
<td><strong>7,061</strong></td>
<td><strong>7,270</strong></td>
</tr>
</tbody>
</table>
NINDS Repository Collections

Genetic Repository
- > 65,000 DNA
- > 60 DNA Panels
- > 11,000 LCL

Biomarker Repository
- > 1000 Subjects
- > 19,000 Plasma
- > 300 Serum
- > 200 CSF
- > 4,500 Urine
- > 2,200 Whole-blood
- > 300 RNA
- > 60 DNA

iPSC/Fibroblasts Repository
- > 40 iPSC
- > 120 Fibroblasts
NINDS Biomarkers Discovery Collection

• PDBP
  – Parkinson’s Disease Biomarkers Program

  5 yrs. 1,400 unique subjects
  Longitudinal Visits 2/yr.
  (900 affected / 500 control)

• BioFIND
  – Fox Investigation for the New Discovery of Biomarkers

  2 yrs. 240 unique subjects
  2 Visits within 2 weeks
  (120 affected / 120 control)

• PREDICT-HD
  – Neurobiological Predictors of Huntington’s Disease

  10+ yrs. 1,380 unique subjects
  Longitudinal Visits 1/yr.
  (230 diagnosed / 850 at-risk / 300 control)
Processing, Quality Control and Assurance

**Incoming Samples**
- Package Arrival
- Whole-Blood
- Plasma
- Serum
- CSF

**Processing**
- Sample Archiving
- DNA & RNA Extraction
- Sub-Aliquoting

**QC & QA**
- Shipment Documentation
- Visual Inspection
- Sample Coding
- Identity & Gender Confirmation
- Freeze / Thaw Tracking
- Hemoglobin Concentration
Integrating Biospecimens with Data
The Role of the Biorepository

**COLLECTION**
- Laboratory Manuals
- Consent Reviews
- Collection Supplies
- Site Training

**DISTRIBUTION**
- Online Display
- Data Access
- Request Review
- Shipment

**PROCESSING**
- Sample Accessioning
- Laboratory Processing
- Quality Control
- Data Sharing
Summary

• The NINDS Repository
  – Publicly accessible
  – Biological specimens and associated de-identified clinical data
  – Sample types
    » DNA, LCL, Fibroblasts, iPSC
    » RNA, Plasma, Serum, CSF, Urine, Whole-blood

• NINDS Biomarker Studies
  – Standardization + Adaptability
    » Sample Collection
    » Laboratory processing, QC & QA
    » Biospecimens and Data Integration
Acknowledgements

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