The PhenX Toolkit

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Standard Measures Needed – c2006

- To detect loci with moderate effect size and G x G; G x E interactions, **large sample sizes needed**.

- Standard (common) measures facilitates **replication of study findings**.

- Potential for **cross-study analysis** increases with common measures

Type 2 Diabetes GWAS (>380K SNPs)
The PhenX Toolkit

• Catalog of recommended measurement protocols
• Facilitate cross-study analysis, increasing impact of individual studies
• Funding: NHGRI + 7 NIH ICs
• PI: Carol Hamilton, RTI International
Content

- 863 Protocols from 28 Research Domains

- Domain Examples:
  - Demographics, Substance Use, Physical Activity
  - Reproductive Health, Pregnancy, Pediatric Development, Geriatrics
  - Diabetes, Cardiovascular, Oral Health

- Special Collections:
Building Consensus

- Steering Committee
- NIH Institutes and Centers
- Expert Working Groups
- Outreach
- Expert Review Panels
Criteria for Protocol Selection

- Standard protocol exists
- Broadly applicable
- Low burden
- Valid & reproducible
- Open source
### Search Results

with filters:

- **Data Collection Mode**
  - Bioassay (3)
  - Clinical Examination (3)
  - Interviewer-administered questionnaire (19)
  - Medical records abstraction (0)
  - Physical Measurement (1)

- **Lifestyle**
  - Infant (2)
  - Toddler (1)
  - Child (6)
  - Adolescent (15)
  - Adult (26)

- **Time to Complete**
  - ≤15 minutes (33)
  - >15 minutes (3)

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**Showing 1 to 36 of 36 protocols**

<table>
<thead>
<tr>
<th>PhenX Protocol</th>
<th>Domain/Collection</th>
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<tbody>
<tr>
<td><strong>Cigarette Nicotine Dependence</strong></td>
<td>Alcohol, Tobacco and Other Substances</td>
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<tr>
<td>Fagerstrom Test for Nicotine Dependence</td>
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<tr>
<td><strong>Substances - Lifetime Abuse and Dependence</strong></td>
<td>Alcohol, Tobacco and Other Substances</td>
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<tr>
<td>National Epidemiologic Survey on Alcohol and Related Conditions-III (NESARC-III)</td>
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<tr>
<td><strong>Withdrawal from Tobacco Use</strong></td>
<td>Tobacco Regulatory Research</td>
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<td><strong>Standardized Tobacco Assessment for Retail Settings</strong></td>
<td>Tobacco Regulatory Research</td>
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<td><strong>Electronic Nicotine Product Device Type</strong></td>
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<tr>
<td>Population Assessment of Tobacco and Health (PATH) Study: Wave 3</td>
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<tr>
<td><strong>Alcohol - Lifetime Use Disorder</strong></td>
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Current Every-Day or Current Some-Day Smokers: Instructions to interviewer: The following six questions are asked twice: once with the lead-in statement about current usage and, if appropriate, a second time with the lead-in statement about the period of maximum usage. a. Currently... (ask 6 questions) b. Was there a 6-month period when you smoked more than you do now? (If "Yes," then continue to ask 6 questions again. If "No," stop.)

1. How soon after you wake up do/did you smoke your first cigarette?
   1. [ ] Within 5 minutes [3 points]
   2. [ ] 6-30 minutes [2 points]
   3. [ ] 31-60 minutes [1 point]
   4. [ ] After 60 minutes [0 points] 2. Do/Did you find it difficult to refrain from smoking in places where it is forbidden, e.g., in church, at the library, in a cinema, etc.?
   1. [ ] Yes [1 point]
   2. [ ] No [0 points]

3. Which cigarette would you hate most to give up?
   1. [ ] The first one in the morning [1 point]
   2. [ ] All others [0 points]

4. How many cigarettes per day do/did you smoke?
   1. [ ] 10 or less [0 points]
   2. [ ] 11-20 [1 point]
   3. [ ] 21-30 [2 points]
   4. [ ] 31 or more [3 points]

5. Do/did you smoke more frequently during the first hours after waking than during the rest of the day?
   1. [ ] Yes [1 point]
   2. [ ] No [0 points]
Linking PhenX and dbGaP Variables for Data Harmonization and Cross-Study Analysis

- PhenX links for 13,655 dbGaP variables, accessible at:
  - dbGaP - a CDE facet browser and a search filter
  - PhenX Toolkit – a search tool and links at variable page
Genomic Medicine Implementation

Scope:

• Understanding of baseline genetic information and their implications

• Impact of genomic interventions on patients, families, and providers

• Assessment of organizational characteristics and readiness to adopt genetic services

Co-chairs: Wendy Chung, MD PhD & Kyle Brothers, MD PhD
Social Determinants of Health

- Individual
  - Discrimination, Food Insecurity, Spirituality
- Structural
  - Concentrated poverty, Social vulnerability

Aims to understand factors that have led to disproportionate burden of the COVID19 pandemic on the underserved and/or vulnerable populations

Co-chairs: Barbara Entwisle, PhD & Alicia Fernandez, MD
COVID-19 Research Collection

Funding: Office of Behavioral and Social Sciences Research (OBSSR); National Institute on Drug Abuse (NIDA); National Institute on Minority Health and Health Disparities (NIMHD); National Cancer Institute (NCI)

• Behaviors and Risks
• History, Treatment and Outcome
• Information Resources
• Psychosocial and Mental Health
• Race, Ethnicity and Demographics
• Socioeconomic
Toolkit Use & Statistics

Top 10 Protocols

1. Ethnicity and Race
2. Current Employment Status
3. Current Age
4. Educational Attainment - Individual
5. Annual Family Income
6. Gender Identity
7. Access to Health Services
8. Food Insecurity
9. Health Literacy
10. English Proficiency

NIH Funding Opportunity Announcements Reference PhenX

394 page visits & nearly 7,000 report downloads

1.6M page visits & nearly 7,000 report downloads

Graph showing the total registered users and total reports downloaded from January 2014 to January 2021.
Acknowledgements

Steering Committee

- Mary Marazita, co-Chair
- Catherine McCarty, co-Chair
- Marylyn Ritchie
- Lindsay Farrer
- Sharon Terry
- Elaine Faustman
- Marsha Treadwell
- Jonathan Haines
- Rosalind Wright

NHGRI

- Jyoti Dayal
- Madison Goldrich
- Teri Manolio

https://www.phenxtoolkit.org/about/teams