Ontology-supported searches for the PhenX Toolkit: Development of a Smart Query Tool (SQT)

Josh Levy, Ying Qin, Huaqin Pan, Carol M. Hamilton
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Welcome to the PhenX Toolkit

The Toolkit provides standard measures related to complex diseases, phenotypic traits and environmental exposures. Use of PhenX measures facilitates combining data from a variety of studies, and makes it easy for investigators to expand a study design beyond the primary research focus. All Toolkit content is available to the public at no cost.

Information about the project is available at www.phenx.org

More →

Please Read Toolkit Guidance

How to cite use of PhenX measures:

Measures incorporated in this study were selected from the PhenX Toolkit version April 29 2013, Ver 5.4. More →

How to cite the PhenX Toolkit:


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PhenX Toolkit Release Notes

Please take a quick survey to tell us how we can improve the Toolkit.

There are a total of 339 measures in the PhenX Toolkit.

Top 5 Measures - Release 5.4
1. Current Age
2. Gender
3. Ethnicity
4. Race
5. Weight

View Top 20 Measures

Top 5 Domains - Release 5.4
1. Demographics
2. Anthropometrics
3. Alcohol, Tobacco and Other Substances
4. Cardiovascular
5. Environmental Exposures
PhenX Domains

- Alcohol, Tobacco, and Other Substances
- Anthropometrics
- Cancer
- Cardiovascular
- Demographics
- Diabetes
- Environmental Exposures
- Gastrointestinal
- Infectious Diseases and Immunity
- Neurology

- Nutrition and Dietary Supplements
- Ocular
- Oral Health
- Physical Activity and Physical Fitness
- Psychiatric
- Psychosocial
- Reproductive Health
- Respiratory
- Skin, Bone, Muscle, and Joint
- Social Environments
- Speech and Hearing
PhenX: consensus measures for Phenotypes and eXposures

• Goal: To identify and disseminate broadly relevant, easily implemented, standard measures of phenotypes and exposures suitable for use in genome-wide association studies (GWAS) and other human subjects research

• The PhenX project is led by RTI International and funded by the National Human Genome Research Institute (NHGRI)
  – RTI International Principal Investigator: Dr. Carol Hamilton
  – NHGRI Project Scientist: Dr. Erin Ramos
PhenX Definitions

• **DOMAIN**: Topical area with a unifying theme
  Alcohol, Tobacco and Other Substances

• **MEASURE**: A certain characteristic of, or related to a study subject
  Tobacco - Nicotine Dependence

• **PROTOCOL**: Standard procedure recommended by a Working Group for collecting a PhenX measure
  Fagerstrom Test for Nicotine Dependence
Challenges

• What can we do to make it easier for Toolkit users to find measures of interest?
• What can we do to more fully support data interoperability?
PhenX and Existing Standards

PhenX Toolkit

dbGaP

CDE Browser

Unified Medical Language System® (UMLS®)

PHIN Vocabulary Access and Distribution System (VADS)

LOINC®

BioPortal
The PhenX SQT - ontology backend

- NCI Metathesaurus (NCIIm)
  - Excellent collection of aliases
  - Well-developed concept associations
  - Based on UMLS semantic network
    - *Semantic network is not an ontology*
Working with NCIm

• Eliminating cycles
  – UMLS and NCIm **Semantic networks** contain cycles, which break a recursive traversal search
  – We use strategies to break these cycles
    • Visited nodes list
    • Visited paths lists

• Ontology size
  – Need to filter paths that do not end in a PhenX node
  – Recursive path building starting with PhenX measures
Resources

- **www.phenx.org**
  - Provides general information about the PhenX project
  - Register to receive periodic updates via e-mail of the PhenX Newsletter and notification of new surveys

- **www.phenxtoolkit.org**
  - Find PhenX measures for inclusion in your study

- **www.genome.gov/gwastudies/**
  - A catalog of published Genome-Wide Association Studies
    (Hindorff et al. PNAS 2009)
Acknowledgements

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  - Erin Ramos (Project Scientist)
  - Heather Junkins (Project Analyst)
  - Teri Manolio
- **SC Members (12)**
  - Jonathan Haines, Chair
  - Bill Harlan, Vice-Chair
- **WG Chairs / Members**
- **SSP (SAA Scientific Panel)**
  - Kenneth Sher, Chair
  - Kevin Conway, NIDA
- **IC Liaisons**
- **dbGaP**
  - Kim Tryka
  - Mike Feolo
- **LOINC**
  - Clem McDonald
  - Daniel Vreeman
- **eMERGE**
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- **NCBO BioPortal**
  - Trish Whetzel, Mark Musen

- **RTI Team**
  - Carol M. Hamilton (PI)
  - Tabitha Hendershot (Co-Investigator)
  - Joe Pratt (Project Manager)
  - Dana Hancock
  - Jane Hammond
  - Wayne Huggins
  - Dean Jackman
  - Debbie Maiese
  - Destiney Nettles
  - Helen Pan
  - Lisa Strader
  - WG Managers
  - Toolkit team
  - Communications team
  - Logistics team
Benefits of Using Standard Measures

- Enables cross-study analysis
  - Increase statistical confidence in results
  - Identify moderate associations and more complex interactions

- Facilitates validation of findings
  - Replicated in independent populations

- Increases the impact of individual studies
  - Many diseases and conditions share common risk factors
Facilitating Cross-Study Analysis

Select PhenX Measures from the Toolkit

Individual Studies

CVD
Diabetes
Obesity
Future Studies

Combine Study Results

Increased Sample Size + Increased Statistical Power

Detect More Subtle and/or More Complex Gene Associations
PhenX Collaborations

• National Center for Biotechnology Information (NCBI) database of Genotypes and Phenotypes (dbGaP)

• Logical Observation Identifiers Names and Codes (LOINC)

• Cancer Data Standards Registry and Repository (caDSR) Common Data Element (CDE) at cancer Biomedical Informatics Grid (caBIG)

• National Center for Biological Ontologies (NCBO) BioPortal
Other NIH Measure Initiatives

• NIH Toolbox for Assessment of Neurological and Behavioral Function (NIH Toolbox)
  http://www.nihtoolbox.org/Pages/default.aspx

• Patient-Reported Outcomes Measurement Information System (PROMIS®)
  http://www.nihpromis.org/?AspxAutoDetectCookieSupport=1

• Quality of Life in Neurological Disorders (Neuro-QOL)
  http://www.neuroqol.org/default.aspx

• Executive Abilities: Methods and Instruments for Neurobehavioral Evaluation and Research (EXAMINER)
  http://examiner.ucsf.edu/index.htm

• NINDS Common Data Elements
  http://www.commondataelements.ninds.nih.gov/#page=Default

• NIH Common Data Elements Portal
  http://cde.nih.gov/
PhenX SQT Summary

- Search functionality
  - Backend ontology
  - Custom mappings (keyword)
  - Full-text searches
- Browsing via tree
  - PhenX Research Domains
  - Conceptual group
- Flexibility in finding measures
  - Choice to browse or search in different ways accommodates scientists with different backgrounds