

PhenX Measures for Data Sharing, Cross-Study Analysis and Data Interoperability

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November 3, 2010

RTI International is a trade name of Research Triangle Institute

PhenX: consensus measures for Phenotypes and eXposures

- The goal of PhenX is to select and define high priority measures useful for Genome-wide association studies (GWAS) and other large-scale genomic research efforts
- The development of the PhenX Toolkit is envisioned as a way to facilitate broad use of common measures by the research community
- PhenX is funded via a Cooperative Agreement with NHGRI

PhenX Approach

- The approach is to select 15 high-priority measures for each of 21 research domains
 - Domains are selected by the PhenX Steering Committee
 - Measures and protocols are chosen by Working Groups which are assembled for each domain
 - Scientific community has the opportunity to review and comment on initial set of up to 25 measures for each domain
- Selected measures are made available to the research community via the PhenX Toolkit to facilitate future cross-study analysis

Building consensus for standard measures of phenotypes and exposures



- PhenX is a three year project led by RTI International and funded by the National Human Genome Research Institute (NHGRI) to contribute to the integration of genetics and epidemiologic research
- PhenX has prioritized 20 research domains related to complex diseases and environmental exposures
- Consensus building is being used to develop a recommended minimal set of high priority measures for use in Genome-wide Association Studies (GWAS) and other large-scale genomic research efforts
- High priority measures will maximize benefits of future research by enabling cross-study comparisons and analysis
- Selection and specification of the measures are driven by the scientific community via the PhenX Steering Committee, Working Groups and community consensus.
- The PhenX Toolkit presents the selected high priority measures to the scientific community

[More...](#)



PHENX TOOLKIT

The PhenX Toolkit makes the results of the project readily accessible via The Internet and enable researchers to implement the high priority measures.

The PhenX Toolkit is now available at: www.phenxtoolkit.org

[More...](#)



STEERING COMMITTEE

A Steering Committee of distinguished experts from the scientific community guides the selection of the measures and promote their use. Research domains include diseases and conditions; lifestyle factors and anthropometrics; and environmental exposures.

[More...](#)



WORKING GROUPS

Working Groups (WGs) are assembled for each research domain. Each WG is responsible for selecting a small set of measures and protocols for obtaining the measure. Recommended measures are vetted with the scientific community through a consensus outreach process.

[More...](#) | [How to get involved](#)

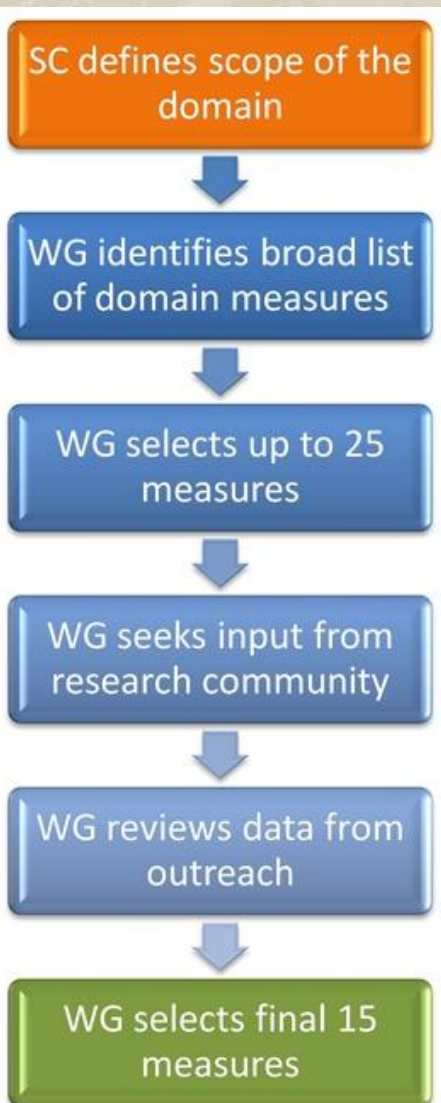
PhenX - Building Consensus

- PhenX Steering Committee (SC)
 - Provides guidance to the project
 - Selects domains and define scope
- NIH Institutes and Centers (IC) Liaisons to SC
 - Provide expertise, outreach
- Expert Working Groups assembled for each domain
 - Refine the scope
 - Select specific measures and protocols
- Outreach to Scientific Community
 - Obtain comments for consideration by the Working Groups

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

Process for Selecting PhenX Measures



- Criteria for measures
 - High quality
 - Low burden for participants and investigators
 - Well established
 - Viable in the future
- Contextually dependent protocols
 - e.g., age- or gender-specific

Welcome to the PhenX Toolkit

Use the Toolkit to browse, search and select PhenX Measures for use in genome-wide association studies (GWAS) or other types of large-scale studies.

For each Measure, the Toolkit has associated protocol(s), references, and links to resources. Use of PhenX Measures will help make your study compatible with other studies that also incorporate PhenX Measures. After selecting PhenX Measures to incorporate in your study, you have the opportunity to generate a Report that provides details on each of the selected PhenX Measures and how they can be incorporated into your study.

PhenX Measures are selected by [Working Groups \(WG\)](#) of domain experts using a consensus-based process. During the selection process, the WGs are asked to consider a number of criteria that were defined by the PhenX Steering Committee (SC). These criteria include that the measures are well established, are high quality and are low burden for investigators and participants. There is a preference for open-source software and nonproprietary instruments.

For more information about PhenX, please visit www.phenx.org.

Please Read Toolkit Guidance

Please cite use of the PhenX Toolkit as:

Measures from the PhenX Toolkit version February 26 2010, Ver 3.0
(www.phenxtoolkit.org) were included in this analysis. PhenX (consensus measures of Phenotypes and eXposures) is supported by NHGRI award No. U01 HG004597

PhenX Toolkit Release Notes

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

Domain

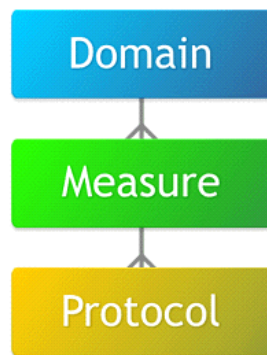
A PhenX Domain is a field of research with a unifying theme and easily enumerated quantitative and qualitative Measures.

Measure

A PhenX Measure refers broadly to a standardized way of capturing data on a certain characteristic of, or relating to a study subject.

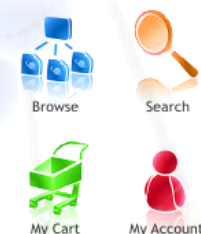
Protocol

A PhenX Protocol is a standard procedure recommended by a Working Group for investigators to collect and record a PhenX Measure.



Quick Start

Tutorial



Registration

You may [browse](#) the PhenX Toolkit, but to save your work, you need to [Register](#).

Existing users may login:

User ID:

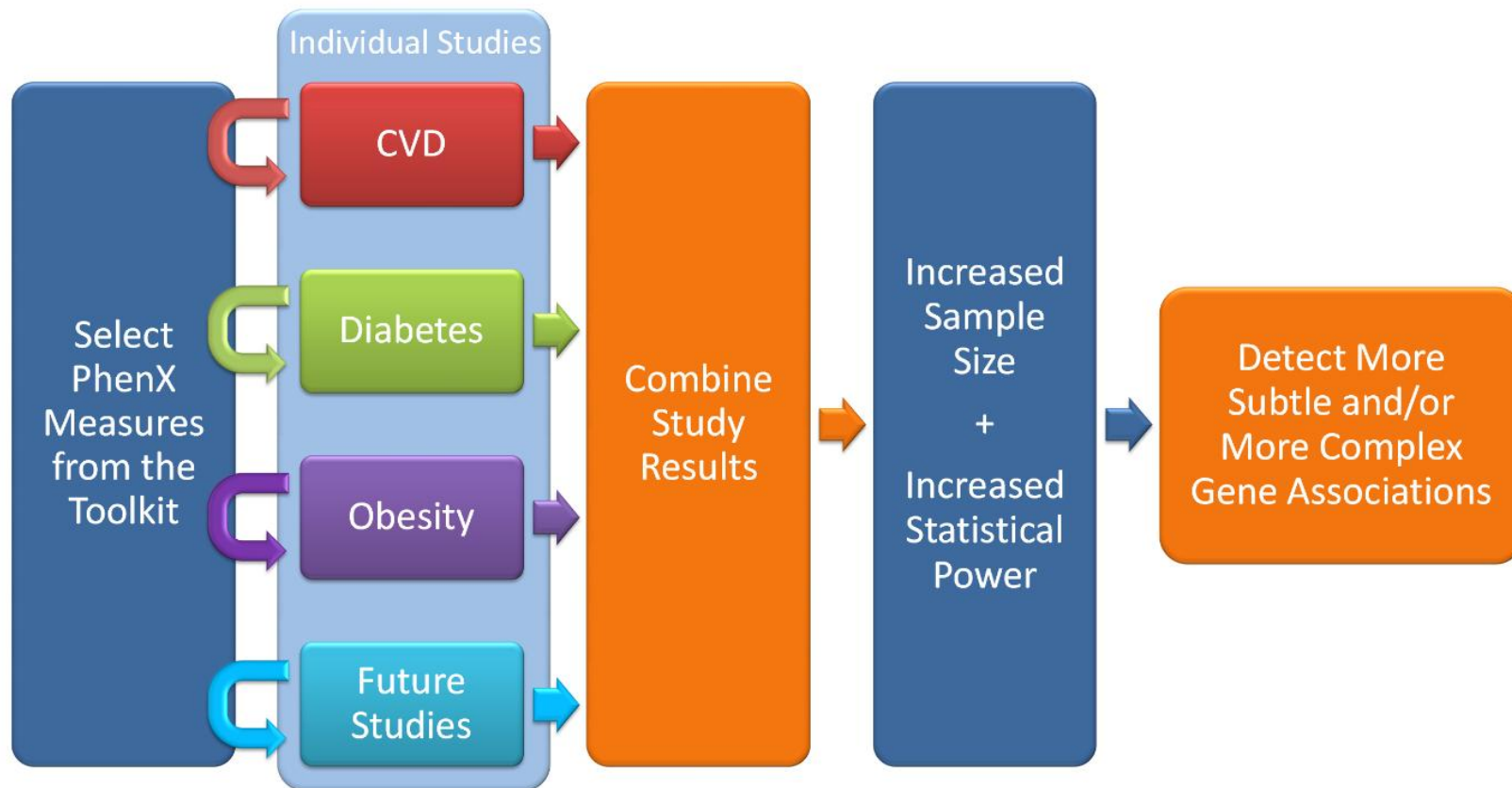
Password:



PhenX Toolkit

- Researchers will visit the Toolkit to:
 - Add standard measures to ongoing studies
 - Consider PhenX measures when planning new studies
 - Obtain high quality measures outside of their area of expertise
 - Review PhenX measures that relate to their primary research focus
- By selecting some of the PhenX measures:
 - Ensure that their study will be compatible with others that also incorporate PhenX measures
 - Combine studies to increase statistical power and the ability to identify genes associated with complex diseases
 - Potential to cumulatively combine with future studies

Facilitating Cross-Study Analysis



What the PhenX Toolkit Is

What the PhenX Toolkit is

- a catalog of recommended measures for inclusion in new studies or when expanding existing studies
- a database that allows researchers to browse, search, and select measures
- cross-referenced to Cancer Biomedical Informatics Grid Common Data Elements
- freely available to the scientific community

What the PhenX Toolkit is not

- not a new set of standards
- not a new ontology of phenotypes
- not a data repository
- not a biobank
- not restrictive
- not a proprietary resource

PhenX Toolkit Contents

- Recommended measures for each research domain
- Detailed protocols for collecting the measures
- Methods to harmonize measures – as needed
- Information about the measures
 - Rationale for inclusion
 - References
- User support
 - User's Manual, Data Dictionary, and Glossary of Terms
 - Quick Start Guide
 - Frequently Asked Questions (FAQs)
 - Derived Variables
 - Cancer Bioinformatics Grid (caBIG) Common Data Elements (CDEs)
 - Supplemental Information

Toolkit Features

- Users can Search or Browse Measures
 - Browse by domain or by conceptual group
 - Search using “Smart Query Tool”
- Users choose Measures and add them to a to Cart
 - Alerts Users to add “essential measures” (additional PhenX measures that are needed to effectively interpret the data)
 - Recommends related measures
 - Registration allows users to save and share their Cart(s)
- Users can generate Reports
 - Download, review, print or save
 - Registered users can add notes to measures, and protocols
- Integrating PhenX Measures
 - Data Collection Worksheet
 - Data Dictionary (variable names and characteristics)



Protocol Overview

[Top](#) » [Physical Activity and Physical Fitness](#) » [Cardiorespiratory Fitness](#) » [Cardiorespiratory Fitness-Exercise Test Estimate \[One Mile Walk\]](#)

Note: Some Protocols contain images. You may click the thumbnails to preview the full image. To print Protocols with full size images, please add those Protocols to the [Cart](#) and [Generate a Report](#).

CARDIORESPIRATORY FITNESS-EXERCISE TEST ESTIMATE [ONE MILE WALK] #150101

Jump to section:



[Show Summary](#)

Description of Protocol

The participant is asked to complete a one-mile walk test as quickly as possible on a track. A technician utilizes a stopwatch to time the walk. The participant wears a heart rate monitor that records h/her heart rate continuously through the test. After the test is completed the technician records the heart rate from the last 10 seconds of the test. In addition, the participant's age, sex, and body weight are needed to complete an equation to determine maximal oxygen consumption (VO_{2max}).

Specific Instructions

Protocol Text

Track or pre-measured one-mile distance

Note: if a track is not used, the location where the timed walk takes place must be flat and pre-measured to equal 1 mile.

First, obtain the participant's weight in pounds via a reliable scale, and also record his/her sex, and current age. Secure the HR monitor chest strap and watch to the participant. Explain that you will time the participant as he/she walks as quickly as possible for one-mile.

Once the participant is ready at the starting line, say "go" and start the stopwatch. Time the participant until he/she completes

Protocol Overview

[Top](#) » [Physical Activity and Physical Fitness](#) » [Cardiorespiratory Fitness](#) » [Cardiorespiratory Fitness-Exercise Test Estimate \[One Mile Walk\]](#)

Note: Some Protocols contain images. You may click the thumbnails to preview the full image. To print Protocols with full size images, please add those Protocols to the [Cart](#) and [Generate a Report](#).

CARDIORESPIRATORY FITNESS-EXERCISE TEST ESTIMATE [ONE MILE WALK] #150101

Jump to section:

Show Summary

Jump to section:

Description of Protocol

Special Instructions

Protocol Text

Selection Rationale

Source

Personnel and Training Required

Equipment Needs

Common Data Elements

General References

Protocol Type

Lifestage

Derived Variables

Requirements

Perform a one-mile walk test as quickly as possible on a track. A technician utilizes a stopwatch to time the participant. A heart rate monitor that records h/her heart rate continuously through the test. After the test, the technician records the heart rate from the last 10 seconds of the test. In addition, the participant's age, sex, and weight are used to complete an equation to determine maximal oxygen consumption (VO_{2max}).

Distance

The location where the timed walk takes place must be flat and pre-measured to equal 1 mile.

First, obtain the participant's weight in pounds via a reliable scale, and also record his/her sex, and current age. Secure the HR monitor chest strap and watch to the participant. Explain that you will time the participant as he/she walks as quickly as possible for one-mile.

Once the participant is ready at the starting line, say "go" and start the stopwatch. Time the participant until he/she completes

Smart Query Tool

Enter search term or PhenX ID:

Show results per page

[Smart Search](#)[Text Search](#)

Smart Search:

- Searches through names, aliases, and keywords; high-specificity
- Returns fewer results than the Text Search, but they are more specific

Text Search:

- Searches through full text of measures and protocols; high-sensitivity, low-specificity
- Returns more results than the Smart Search, but they are less specific
- Also searches through Supplemental Information

Helpful Search Tips:

- Use "quotation marks" to search for exact phrase
- For high specificity, search with a single key term. For example, to find "Family history of diabetes among first degree relatives", search "family history" (with quotes)

[See search logic](#) for more details

Data Sharing and Data Interoperability

Collaborations:

- National Library of Medicine (NLM)
 - 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)
- Public Population Project in Genomics (*P3G*) Data Schema and Harmonization Platform for Epidemiological Research (DataSHaPER)
- electronic Medical Records & Genomics (eMERGE) network
- National Institute of Neurological Disorders and Stroke (NINDS)

Collaboration with dbGaP

- Highlight PhenX measure for variables in dbGaP



Variable mapped to PhenX measure can be flagged

Clinical Variable	Dataset	Variable Description	Variable ID
HBP_SBHS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa02L1_2_trans GenADA CSA02I1 Medical History	HIGH BLOOD PRESSURE: Subject History of disease	phv00081405.v1.p1
HBP_FAMHS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa02L1_2_trans GenADA CSA02I1 Medical History	HIGH BLOOD PRESSURE: Family History of disease	phv00081406.v1.p1
HBP_FAMNO GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa02L1_2_trans GenADA CSA02I1 Medical History	HIGH BLOOD PRESSURE: Number of family members with history of disease	phv00081407.v1.p1
HBP_SBHS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa04L1_trans GenADA CSA04I1 Medical History	HIGH BLOOD PRESSURE: Subject History of disease	phv00081536.v1.p1
HBP_FAMHS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa04L1_trans GenADA CSA04I1 Medical History	HIGH BLOOD PRESSURE: Family History of disease	phv00081537.v1.p1
HBP_FAMNO GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa04L1_trans GenADA CSA04I1 Medical History	HIGH BLOOD PRESSURE: Number of family members with history of disease	phv00081538.v1.p1
HBP_SBHS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa06_trans GenADA CSA06 Medical History	HIGH BLOOD PRESSURE: Subject history of disease	phv00081683.v1.p1
HBP_FAMHS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa06_trans GenADA CSA06 Medical History	HIGH BLOOD PRESSURE: Family history of disease	phv00081684.v1.p1
HBP_FAMNO GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_csa06_trans GenADA CSA06 Medical History	HIGH BLOOD PRESSURE: Number of family members with history of disease	phv00081685.v1.p1
BPD GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_vitl GenADA Vital Signs - Measurements	Diastolic Blood Pressure	phv00082599.v1.p1
BPS GenADA/LONG/Imaging (Genetic Alzheimer's Disease Associations)	d_vitl GenADA Vital Signs - Measurements	Systolic Blood Pressure	phv00082600.v1.p1

LOINC Codes for PhenX Measures



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- Meetings
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Logical Observation Identifiers Names and Codes (LOINC®)



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The purpose of LOINC® is to facilitate the exchange and pooling of clinical results for clinical care, outcomes management, and research by providing a set of universal codes and names to identify laboratory and other clinical observations. The [Regenstrief Institute, Inc](#), an internationally renowned healthcare and informatics research organization, maintains the LOINC database and supporting documentation, and the RELMA mapping program.

LOINC News

Current Versions


LOINC 2.32
Released: 2010-06-30

RELMA 4.5
Released: 2010-06-30

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2010-06-30

 [LOINC Version 2.32](#)
and [RELMA Version 4.5](#)
Available
2010-06-30

LOINC Codes for PhenX Measures



LOINC	Component	Property	System	Time	Scale	Method	Class	Type	Status	Short name
58243-7	Angina protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Angina protocol
56054-0	Arm Span Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Arm Span Proto
58248-6	Arrhythmia protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Arrhythmia prot
58343-5	Cardiovascular protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Cardiovascular p
56049-0	Current Age Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Current Age Pro
56050-8	Ethnicity Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Ethnicity Protoc
56059-9	Hand Dominance Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Hand Dominanc
56060-7	Height Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Height Protocol
58245-2	Myocardial infarction protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Myocardial infar
58251-0	Rheumatic fever protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Rheumatic feve
56084-7	Weight Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Weight Protoco
58247-8	Abdominal aortic aneurysm protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Abd aneurysm p
56055-7	Birth Weight Protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Birth Weight Pro
58240-3	Blood pressure protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Blood pressure
58249-4	Deep venous thrombosis protocols	-	^Patient	Pt	-	PhenX	PANEL.PHENX	clinical	TRIAL	Deep venous th PhenX

PhenX CDEs in caBIG



CDE Browser

[Admin Tool](#)

[Curation Tool](#)

[NCI Metathesaurus](#)

[NCI Terminology Server](#)

[Sentinel Tool](#)

[UML Model Browser](#)

Search for Data Elements

15 Matches

caDSR Contexts>>caBIG (NCI cancer Biomedical Informatics Grid)>>Classifications>>PhenX>>ATOS (Alcohol, Tobacco, and Other Substances)

- ☒ Exact phrase
- ☐ All of the words
- ☐ At least one of the words

Tip: This is an exact match search. To search for partial words or phrases use the * as a wildcard.

Note: Default settings exclude Test and Training Context views from the tree and certain 'non-released' Workflow and Data Element views. To view or change the exclusion criteria, Search Preferences' will be reset to default settings when the 'New Search' button is clicked.

Search

Clear

New Search

Search Results [Search within results](#)

Results fewer than expected? Check Search Preferences

[\[Download Data Elements to Prior Excel\]](#)

[\[Download Data Elements to Excel\]](#)

[\[Download Data Elements to PDF\]](#)

[\[Download CDE Browser DT\]](#)

Sort order : (Default) Registration Status>>Workflow Status>>Long Name [Ascending]

[Add to CDE Cart](#)

[Add to CDE compare list](#)

[Compare CDEs](#)

<input type="checkbox"/> Long Name	Preferred Question Text	Owned By
<input type="checkbox"/> Alcohol 24 hours	During the periods when you drank the most, what was the LARGEST number of drinks that you drank in a single day?	caBIG
<input type="checkbox"/> Maximum Consumption Number		



Cross-reference table for PhenX measures and variables

PhenX variable	PhenX ID	dbGaP variable	LOINC code	P3G variable*	eMERGE variable	caBIG CDE
Current Age	PXV010101020000	phv00023938.v1 phv00024004.v1	21612-7	Age	Age	2423393
Ethnicity	PXV010501010000	phv00023941.v1 phv00024007.v1	56050-8	Ethnicity	Ethnicity	2200284
Gender	PXV010701010000	phv00023939.v1 phv00024005.v1	46607-8	Gender	Gender	2179640

* P3G DataShaPER doesn't distinguish the "exact" and "similar" mappings to the PhenX measures

RTI International is a trade name of Research Triangle Institute

Toolkit Use Summary

(www.phenxtoolkit.org – Month End September 2010)

Number of Visits: 126,915

Number of Unique Visits: 16,155
(by unique ip address)

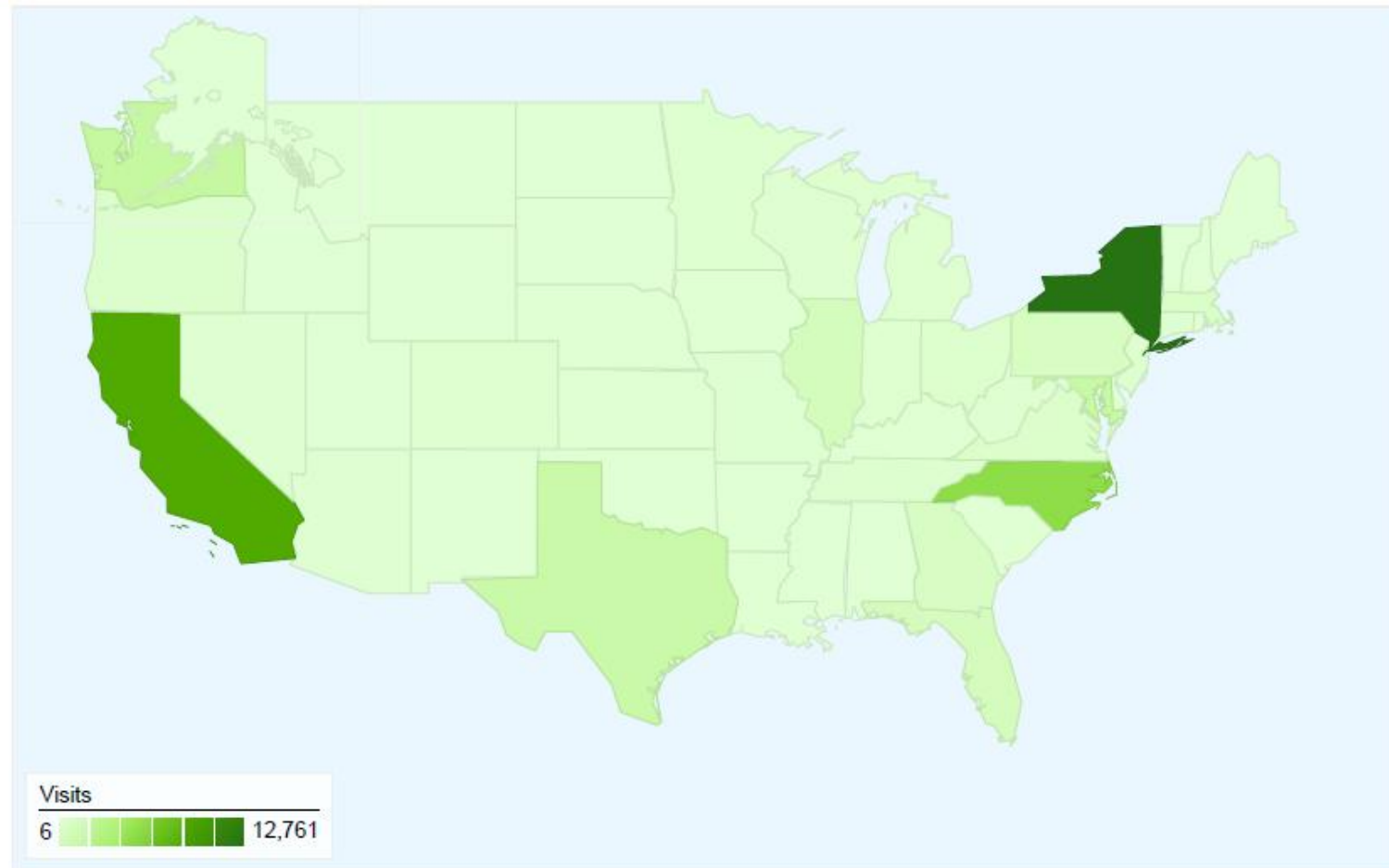
Total Page Views: 378,761

Average Number Visits Per Day: 180

Total # of Countries with Toolkit accesses: 117

Number of Registered Users: 315

Visits from the United States



Visits from Top Countries

Rank	Country	No. of Unique Visitors
1	United States	8540
2	United Kingdom	926
3	Australia	565
4	Canada	493
5	India	458
6	Philippines	216
7	Germany	162
8	China	152
9	Brazil	124
10	Malaysia	116
11	Italy	107
12	Netherlands	106
13	New Zealand	83
14	Thailand	80
15	Singapore	76

Resources

- www.phenx.org
 - Register to receive periodic updates via e-mail of the PhenX Newsletter and notification of new surveys
- www.phenxtoolkit.org
 - Additional measures and protocols will be included in the toolkit as they become available
- www.genome.gov/gwastudies/
 - A catalog of published Genome-Wide Association Studies (Hindorff et al. PNAS 2009)

Acknowledgements

- NHGRI
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 - Bill Harlan, Vice-Chair
- WG Chairs / Members
- IC Liaisons
- dbGaP
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 - Jayashri Mehta
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 - Tabitha Hendershot
 - Wayne Huggins
 - Dean Jackman
 - Richard Kwok
 - Debbie Maiese
 - Destiney Nettles
 - Helen Pan
 - WG Managers
 - Toolkit team
 - XC team
 - Communications team
 - Logistics team