

The PhenX Toolkit: Standard Measures for Collaborative Research

Clinical Common Data Elements Task Force
(CDETF)

November 4, 2016

Carol M. Hamilton, PhD

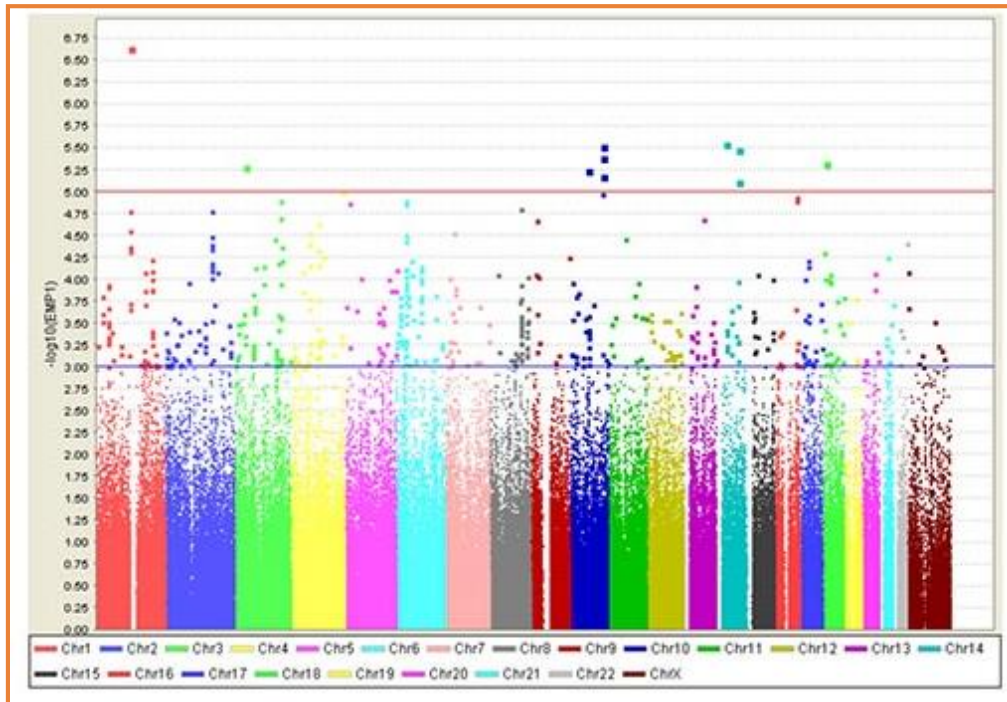
RTI International is a trade name of Research Triangle Institute

PhenX: consensus measures for **Ph**enotypes and **eX**posures

- PhenX is funded by the National Human Genome Research Institute (NHGRI), Project Scientist, Dr. Erin Ramos with co-funding from the National Institute on Drug Abuse (NIDA)
- In September 2007, RTI International was awarded a Cooperative Agreement (U01) to select and define high priority measures for GWAS (phase 1)
- In July 2013, RTI International was awarded a Genomics Resource grant (U41) to expand the Toolkit, review existing measure and add features (phase 2)
- PhenX uses an established consensus based process, and is driven by the scientific community
- Initially focused on measures for genome-wide association studies, the scope has broadened to clinical, translational and epidemiological research

Standard Measures Needed – c2006

Type 2 Diabetes GWAS (>380K SNPs)



(www.broad.mit.edu/diabetes/scandinavs/type2.html)

- Combining studies increases ability to detect loci with moderate effect size ($G \times G$; $G \times E$ interactions)
- Replication of GWAS results
- Potential for cross-study analysis and replication limited by lack of standardized measures

The PhenX Project

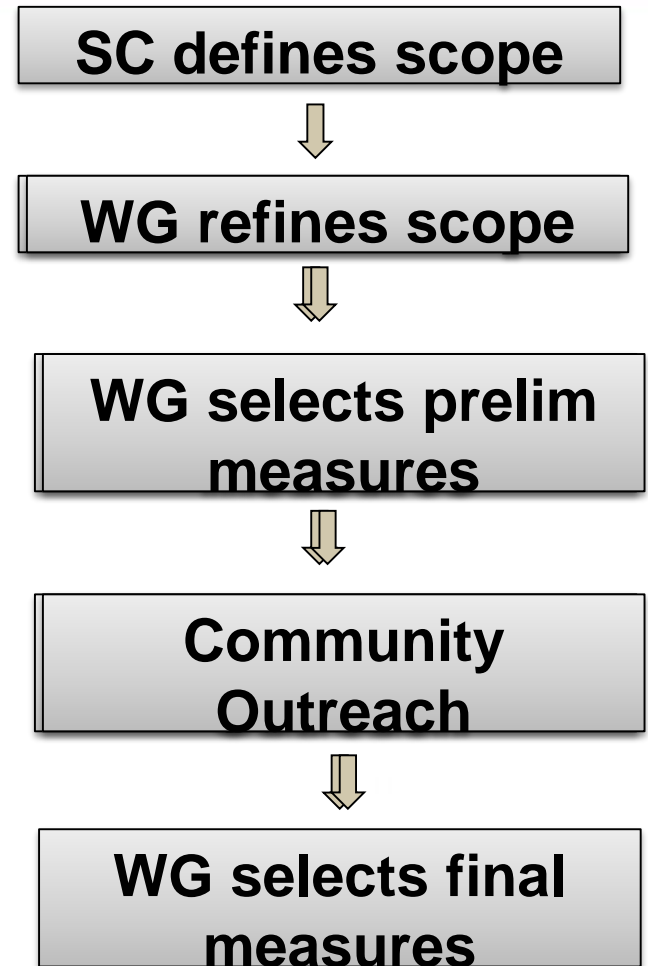
- **Goal:** Produce an online resource of standard phenotypic & environmental exposure measures
- Cooperative Agreement (2007 U01 → 2013 U41)
- ~ 450 measures addressing 21 domains
- Protocols, data element dictionaries, data collections worksheets



(www.phenxtoolkit.org)

Selecting PhenX Measures

- Criteria for selecting measures include:
 - Well-established and broadly validated
 - Low burden to participants & investigators
 - Applicable across population groups
 - Freely available protocols (preferred)
 - Useful to investigators who are *not* domain experts



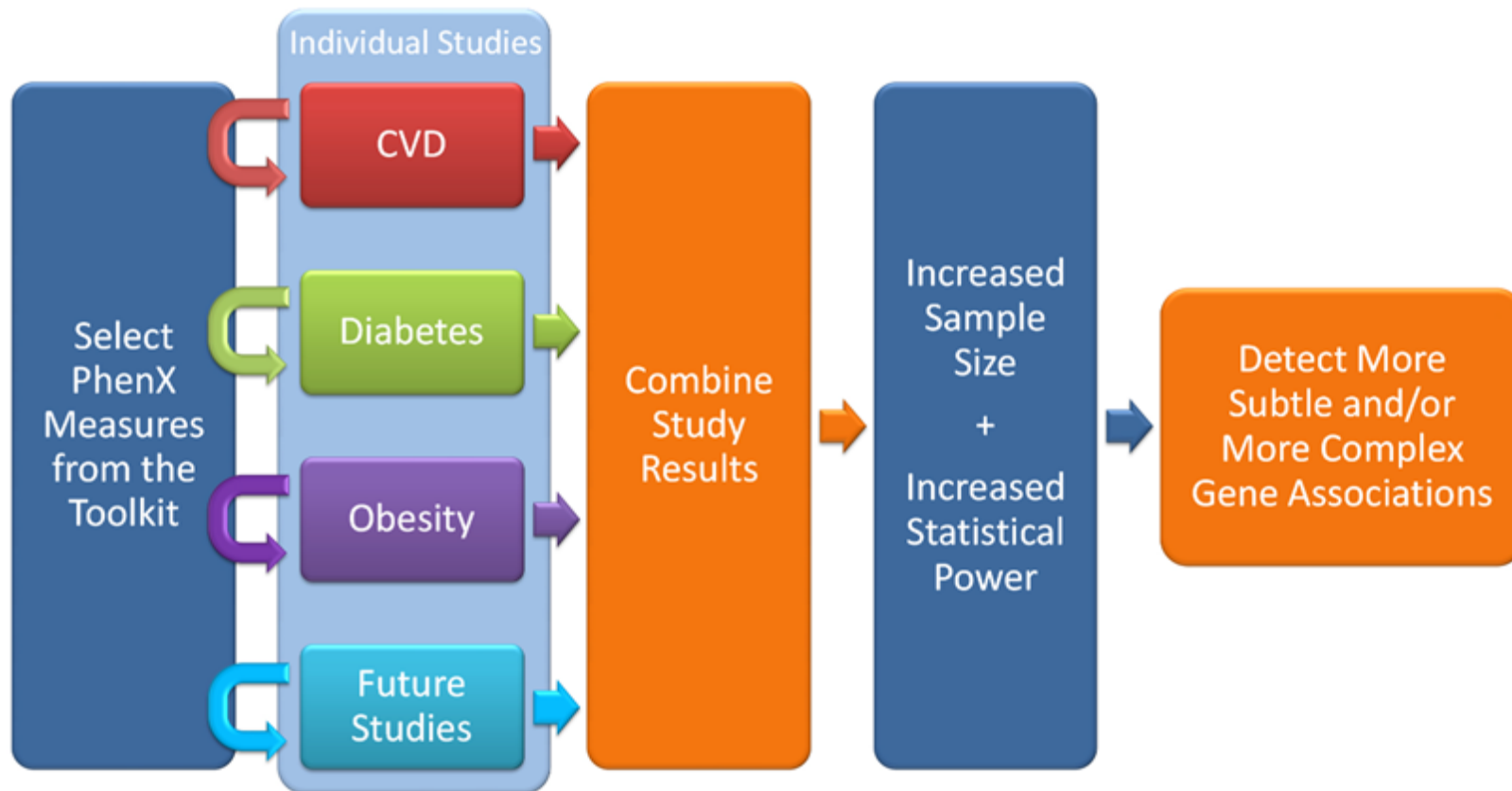
PhenX Project Organization



Why Use Standard Measures?

- Study findings require validation
 - Initial findings need to be replicated, standard measures aid comparisons
- Increased sample size provides greater statistical power
 - For GWAS, to identify moderate associations and more complex interactions
 - For other types of studies, increases statistical confidence in results
- Cross-study analyses increase the impact of individual studies
 - Many diseases and conditions share common risk factors
 - Use of standard (common) measures facilitates cross-study analyses

PhenX Measures Enable Cross-Study Analysis



PhenX Terminology

- **DOMAIN:** Topical area with a unifying theme
Sickle Cell Disease Neurology, Quality of Life, and Health Services
- **MEASURE:** A certain characteristic of, or related to, a study subject
Quality of Life in Sickle Cell Disease
- **PROTOCOL:** Standard procedure recommended by a Working Group to collect and record a PhenX measure
 - Adult Sickle Cell Quality of Life Measurement Information System (ASCQ-Me)
 - PedsQL Sickle Cell Disease Module



PhenX - Research Domains

- Alcohol, Tobacco, and Other Substances
- Anthropometrics
- Cancer
- Cardiovascular
- Demographics
- Diabetes
- Environmental Exposures
- Gastrointestinal
- **Geriatrics**
- Infectious Diseases and Immunity
- Neurology
- Nutrition and Dietary Supplements
- **Obesity**
- Ocular
- Oral Health
- Physical Activity and Physical Fitness
- **Pregnancy**
- Psychiatric
- Psychosocial
- **Rare Genetic Conditions**
- Reproductive Health
- Respiratory
- Skin, Bone, Muscle, and Joint
- Social Environments
- Speech and Hearing

Criteria for Selecting PhenX Measures

- Clearly defined
 - Well established
 - Broadly applicable
 - Validated
 - Reproducible
 - Specific
 - Reliable
 - Standard measurement protocols exist
-
- Criteria defined by the PhenX Steering Committee
 - Working Groups select measures to be included in the Toolkit

PhenX Supplements - adding depth to the Toolkit

- PhenX Measures for Sickle Cell Disease Research (funded by NHLBI)
 - Ellen Werner, Project Scientist
 - Sickle Cell Disease Research and Scientific Panel (SRSP)
 - Collaborating with H3Africa (Human Heredity and Health in Africa) on a Sickle Cell Disease ontology
- PhenX Measures for Mental Health Research (funded by NIMH)
 - Greg Farber, Project Scientist
 - Mental Health Research Scientific Panel (MHRP)
- PhenX Measures for Tobacco Regulatory Science (funded by TRSP)
 - Kay Wanke, Project Scientist
 - Tobacco Regulatory Research Scientific Panel (TRRP)
- PhenX Measures for Substance Abuse and Addiction (funded by NIDA)
 - Kevin Conway, Project Scientist
 - Substance Abuse and Addiction Scientific Panel (SSP)



Welcome to the PhenX Toolkit

The PhenX (consensus measures for **Phen**otypes and **eX**posures) Toolkit is a catalog of recommended, standard measures of phenotypes and environmental exposures for use in biomedical research. PhenX measures can be used to expand a study design beyond the primary research focus. Use of PhenX measures facilitates cross-study analysis, potentially increasing the scientific impact of individual studies. The PhenX Toolkit is a Web-based resource and is available for use at no cost. [More >>](#)



Citation and
Guidance



Research Domains



Register Your
Study



Newsletter



Mental Health
Research



Sickle Cell Disease
Research



Substance Abuse
and Addiction



Tobacco Regulatory
Research

Types of PhenX Measures

[My Toolkit](#)[Register](#) | [Log In](#)[Home](#)[Browse](#)[Search](#)[Registration](#)[Resources](#)[News](#)[Help](#)[About](#)

Smart Query Tool

Search pre-defined search filters listed below

Search by entering your own terms in the box below

Data Collection Mode

- ☒ Interviewer-administered questionnaire
- ☒ Self-administered questionnaire
- ☐ Bioassay
- ☒ Clinical Examination
- ☐ Physical Measurement
- ☐ Medical records abstraction
- ☐ Secondary Data Analysis

Lifestage

- ☐ Infant
- ☐ Toddler
- ☐ Child
- ☐ Adolescent
- ☐ Adult
- ☐ Senior
- ☐ Pregnancy

Time to Complete

- ☐ ≤15 minutes
- ☐ >15 minutes

Language

- ☐ English
- ☐ Spanish
- ☐ Other

Enter search term or PhenX ID:

Show results per page

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

Protocol Overview

☐ Show Tree

[Browse](#) » [Domains](#) » [Diabetes](#) » [Urinary Creatinine Assay for Kidney Function](#) » [Urinary Creatinine Assay for Kidney Function Protocol](#)

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 your Toolkit and [Generate a Report](#).

URINARY CREATININE ASSAY FOR KIDNEY FUNCTION #141601

[Protocol Release Date](#) ⌵[Description of Protocol](#) ⌵[Specific Instructions](#) ⌵[Protocol Text](#) ⌵[Selection Rationale](#) ⌵[Source](#) ⌵[Personnel and Training Required](#) ⌵[Equipment Needs](#) ⌵[Standards](#) ⌵[General References](#) ⌵[Protocol Type](#) ⌵[Derived Variables](#) ⌵[Requirements](#) ⌵

Review Measure

☐ Show Tree

Browse » Domains » Diabetes » Urinary Creatinine Assay for Kidney Function

MEASURE: Urinary Creatinine Assay for Kidney Function #141600

Definition: A bioassay to measure urine concentration of creatinine, a muscle metabolite that is filtered out of blood by the kidneys.

Purpose: The urinary creatinine measure should be done in conjunction with urinary microalbumin to determine ratio of urine albumin to urine creatinine which can predict the risk of nephropathy (National Health and Nutrition Examination Survey and University of Minnesota Laboratory Procedure Manual for Urinary Creatinine, 2008).

Protocols:

[Add to My Toolkit](#) #141601 Urinary Creatinine Assay for Kidney Function »

Essential Measures

?

Related Measures?

Blood Pressure (Adult/Primary)
Personal History of Kidney Failure

Current Age
Gender

Serum Creatinine Assay for Kidney Funct...
Urinary Microalbumin Assay for Kidney F...

Collections?

Organ Function
Kidney Failure

Maternal Complications

Sickle Cell Disease: Cardiovascular, Pulmonary, and Renal Specialty Collection

Keywords: Centers for Disease Control and Prevention, CDC, National Center for Health Statistics, NCHS, National Health and Nutrition Examination Survey Questionnaire, NHANES, University of Minnesota, Fairview University Medical Center, Nephropathy, diabetic nephropathy, kidney, kidney disease, kidney failure, Cardiovascular disease, Microalbuminuria, Macroalbuminuria, Diabetes

Measure Release Date: May 12, 2010



Add this Measure* to My Toolkit



Add this Measure* and all Essential Measures to My Toolkit

PhenX and Data Science

- Customized for each Toolkit
 - Data Collection Worksheets (DCW) and
 - Data Dictionaries (DD)
 - dbGaP compatible format
 - REDCap compatible format
 - Web-based data collection (electronic Case Report Form (eCRF))
- Identify common measures across studies
 - Link Your Study – compare with ongoing studies
 - dbGaP mapping – legacy studies
 - 790 studies 200K variables in dbGaP
 - 636 protocols, 22K variables in PhenX

PhenX Collaborations

- The database of Genotypes and Phenotypes (dbGaP)
- REDCap (Research Electronic Data Capture)
- PhenX in NIH CDE Resources
- H3Africa Sickle Cell Disease Ontology



dbGaP

The database of Genotypes and Phenotypes (dbGaP) was developed to archive and distribute the results of studies that have investigated the interaction of genotype and phenotype.

- Identify database of Genotypes and Phenotypes (dbGaP) studies using PhenX measures for potential use in cross-study analysis
- Investigators can search dbGaP and find studies that included measures comparable or related to PhenX measures
- Mapping PhenX variable to dbGaP study variables retrospectively (PhenX has relatively small number of variables compared to dbGaP)
- In the future, investigators submitting data to dbGaP will be asked to self-identify PhenX measures

dbGaP

dbGaP

alcohol[PhenX]

Create alert

Limits

Advanced

20 per page

Search results

Items: 18

Search results: 218 Variables, 0 Analyses, 0 Documents, and 0 Datasets in 18 Studies

Studies (18)

Variables (218)

Study Documents (0)

Analyses (0)

Datasets (0)

| Study | Embargo Release | Details | Participants | Type Of Study | Links | Platform |
|--|---|---------|--------------|-----------------------|-----------------------|--|
| phs000007.v27.p10 Framingham Cohort | Versions 1-25: passed embargo Version 26: 2016-08-07 Version 27: 2017-01-15 | V D A S | 15172 | Longitudinal | Links | HuGeneFocused50K_Affy Mapping250K_Nsp Mapping250K_Sty Mapping50K_Hind240 Mapping50K_Xba240 |
| phs000763.v1.p1 The Collaborative Study on the Genetics of Alcoholism (COGA) | Version 1: passed embargo | V D A S | 3557 | Family | Links | HumanOmniExpress-12v1_C OmniExpress 1M Duo 1000 Genomes |
| phs000285.v3.p2 CARDIA Cohort | Versions 1-3: passed embargo | V D A S | 3622 | Longitudinal | Links | |
| phs000256.v3.p2 The Vaginal Microbiome: Disease, Genetics and the Environment | Versions 1-3: passed embargo | V D A S | 3474 | Twin, Clinical Cohort | Links | 454 GS FLX Titanium 454 GS FLX Titanium |
| phs000170.v2.p1 GWAS on Cataract and HDL in the PMRP | Version 1: passed embargo Version 2: 2016-03-09 | V D A S | 3989 | Case-Control | Links | Human660W-Quad_v1_A |





20 per page

Search results

Items: 1 to 20 of 218

Search results: 218 Variables, 0 Analyses, 0 Documents, and 0 Datasets in 18 Studies

- Studies (18)
- Variables (218)
- Documents (0)
- Analyses (0)
- Datasets (0)

| Clinical Variable | Dataset | Variable Description | Variable ID |
|---|---|--|------------------------------------|
|  G3A113 Framingham Cohort | ex3_1s Clinic Exam, Generation 3 Exam 1 | IF EVER CONSUMED ALCOHOL: HOW OLD WERE YOU WHEN YOU FIRST STARTED DRINKING ALCOHOLIC BEVERAGE? | phv00020968.v4.p10 |
|  G3A137 Framingham Cohort | ex3_1s Clinic Exam, Generation 3 Exam 1 | IF EVER CONSUMED ALCOHOL: WHAT WAS THE MAXIMUM NUMBER OF DRINKS YOU HAD IN 24 HOUR PERIOD DURING THE PAST MONTH? | phv00020992.v4.p10 |
|  q3b0114 Framingham Cohort | e_exam_2011_m_0017s Clinic Exam: Interview, Physical Exam, ECG, Generation 3, New Offspring Spouse, and Omni 2, Exam 2. The inclusion criteria for this dataset is at least 19 years old at Exam 1, at least one parent in the Framingham Heart Study, signed BUMC IRB consent at exam 1 for participation in the FHS Third Generation exam and participation in FHS genetic research. | Medical History-Alcohol Consumption: What was the maximum number of drinks you had in 24 hr. period during the past month? | phv00177321.v2.p10 |
|  age first drink The Collaborative Study on the Genetics of | COGA Family GWAS Subject Phenotypes The dataset provides data collected for alcohol abuse, dependence (e.g. onset, status at first diagnosis, and presence of maximal symptoms based on information | Age of first whole drink | phv00217597.v1.p1 |

Filter

Al
di
db
S
S

Find
Data

F

Rece

Q a
Q a
Q F
1
Q F
2
Q F
1

Research Electronic Data Capture (REDCap)

- Study design tool for CTSA funded studies
- All PhenX protocols have REDCap Zip Instruments, which can be downloaded then upload directly to REDCap
- Web-based data collection for PhenX protocols
- Accessible at both REDCap (a link to PhenX) and PhenX sites
- REDCap available for use on mobile devices




My Toolkit

[Show Tree](#)[« Return to last visited browse page](#)[Download Report](#)[Download Data Collection Worksheet](#)[Download Data Dictionary](#)[Download !\[\]\(870f5d5e9c0d57485634be3ecf52f3ca_img.jpg\) REDCap Instrument Zip](#) Empty My Toolkit

| Measure | Protocol | Requirements? | Essential Measures? |
|---------|----------|---------------|---------------------|
|---------|----------|---------------|---------------------|


| | | | |
|--------------|--|--|--|
| Birth Weight | Birth Weight - Measured Weight at Birth Remove | | |
|--------------|--|--|--|

[Remove](#) | [Related Measures](#)

 Essential Measures are not in the cart

Review additional Essential Measures:

☐ Ethnicity☐ Gender☐ Race

 Essential Measures are needed to interpret your data. [Add all Essential Measures to your Toolkit »](#)

[Share this Toolkit »](#)



 [Consortium Wiki](#) (Login Required)

[Introduction](#)

[Software](#)

[Consortium Partners](#)

[Become a Partner](#)

[Video Resources](#)

[Citing REDCap](#)

[Library](#)

[REDCapCon](#)

REDCap "Instrument ZIP" files

REDCap 6.5.0 and later provides a way to easily add new data collection instruments to a REDCap project by uploading an "instrument ZIP" file. These instrument ZIP files may be downloaded from an external instrument library (e.g., PhenX Toolkit, Medical Data-Models) or from another REDCap project or user, and then uploaded on the Online Designer page in a REDCap project so that they get added as a new data collection instrument. These ZIP files can contain attachment files for "descriptive" fields, which makes it very easy to obtain or share an instrument containing several attachment files.

NOTE: An instrument ZIP file contains only one instrument that gets **added** to a project, whereas a REDCap data dictionary CSV file contains ALL the instruments in a project and would thus **replace all** instruments in the project when uploaded.



Logged in as **hpan67@yahoo.com**
Log out

My Projects
Project Home
Project Setup

Project status: **Development**

Data Collection [Edit instruments](#)

Record Status Dashboard
- View data collection status of all records
Add / Edit Records
- Create new records or edit/view existing ones

Data Collection Instruments:

Mv First Instrument

SCD-demo1

Project Setup

Online Designer

Data Dictionary

[VIDEO: How to use this page](#)

The Online Designer will allow you to make project modifications to fields and data collection instruments very easily using only your web browser. NOTE: While in development status, all field changes will take effect immediately in real time.

Data Collection Instruments

Add new instrument:

- Create** a new instrument from scratch
- Import** a new instrument from the official [REDCap Shared Library](#)
- Upload** instrument ZIP file from another project/user or [external libraries](#)

| Instrument name | Fields | View PDF | Instrument actions |
|-----------------|--------|----------|--------------------|
|-----------------|--------|----------|--------------------|

Summary Table for NIH CDE Initiatives

This table lists summary information for [NIH CDE Initiatives](#). More information on NIH CDE Initiatives: [Subject Areas](#), [Detailed Summaries](#).

Show entries

Search:

| Link to Homepage | Link to CDEs | Brief Summary | Number of Elements | Studies and Publications | CDE Resource Contact |
|--|-----------------------------|---|--|------------------------------|---|
| Standardized Asthma Outcomes for Clinical Research | Asthma CDEs | The standardized asthma outcomes for clinical research represent recommendations for core (required in future studies), supplemental (to be used according to study aims), and emerging (requiring validation and standardization) outcomes for 7 domains of asthma clinical research outcome measures. Subject Areas More... | 10 (adults), 25 (children) | -- | NHLBI , NIAID |
| Chronic Low Back Pain CDEs | cLBP | Recommended minimum dataset for research on chronic low back pain. Subject Areas More... | 40 | -- | NCCAM |
| Early Detection Research Program | EDRN | CDEs for use in describing samples and data collected as part of cancer biomarker research. Subject Areas More... | 1,600 | Publications | NCI |
| eyeGENE | eyeGENE | As part of eyeGENE, common data elements have been developed for collecting phenotypic data associated with more than 30 inherited ophthalmic diseases. Subject Areas More... | 300+ | Studies | NEI |
| Global Rare Diseases Patient Registry and Data Repository | GRDR | CDEs to facilitate standardized data collection into the GRDR and to assist organizations in establishing rare disease registries that contribute information to GRDR. Subject Areas More... | 70 | Publications | ORDR |
| Quality of Life Outcomes in Neurological Disorders | Neuro-QOL | A core set of quality-of-life questions that address chronic neurologic disorders, plus sets of supplemental questions specific to targeted diseases or subgroups of patients. Subject Areas More... | 500 | Publications | NINDS |
| NIDA Substance Abuse Electronic Health Record Data Elements | NIDA EHR | A set of brief screening and initial assessment tools for substance use disorders (SUDs) for use in general medical settings. Subject Areas More... | 80+ | -- | NIDA |
| NIH Toolbox for Assessment of Neurological and Behavioral Function | NIH Toolbox | An integrated set of tools for measuring cognitive, emotional, motor and sensory function. Subject Areas More... | 4 batteries of tests, each with 5-24 tests | Publications | NIH |
| NINDS Common Data Elements | NINDS CDEs | A core set of data elements for use in NINDS-funded studies, including core and supplementary sets of data elements for use in disease-specific studies. Subject Areas More... | 10,000 unique variables, 550+ instruments | Studies | NINDS |
| Consensus Measures for Phenotypes and eXposures | PhenX | Standard measures related to complex diseases, phenotypic traits and environmental exposures for inclusion in genome-wide association studies (GWAS) and other large-scale genomic and epidemiologic research efforts. Subject Areas More... | 15,000+ variables, 428 protocols | Publications | NHGRI |
| Patient Reported Outcomes Measurement Information System | PROMIS | A system of item banks measuring patient-reported health status for various domains of physical, mental, and social health across clinical populations (i.e. not disease-specific). Subject Areas More... | 50 item banks | Publications | NIAMS |

Showing 1 to 11 of 11 entries

[Jump to top of page](#)

◀ [Previous](#) [Next](#) ▶



[Admin Tool](#) [Curation Tool](#) [NCI Metathesaurus](#) [NCI Terminology Server](#) [Sentinel Tool](#) [UML Model Browser](#) [What's new](#) [Available Downloads](#) [New!](#)

Data Element Search

Search for Data Elements

[Search preferences](#)

[Advanced search](#)

caDSR Contexts

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

Name ▼

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

| | | | | | | | | |
|--------------------------|--|---|-------|-----------|--|----------|---------|-----|
| <input type="checkbox"/> | NIH Stroke Scale Evaluation Best Language Score | Best Language | NINDS | caBIG | | RELEASED | 3210248 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Evaluation Date | Date of NIHSS | NINDS | caBIG | | RELEASED | 3201206 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Evaluation Dysarthria Score | Dysarthria | NINDS | caBIG | | RELEASED | 3210249 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Evaluation Limb Ataxia Score | Limb ataxia | NINDS | caBIG | | RELEASED | 3210246 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Evaluation Motor Arm Right Score | Motor arm right | NINDS | caBIG | | RELEASED | 3204336 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Evaluation | | | | | | | |
| <input type="checkbox"/> | NIH Stroke Scale Level of Consciousness Question Score | LOC questions | NINDS | caBIG | | RELEASED | 3201209 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Level of Consciousness Score | Level of consciousness | NINDS | caBIG | | RELEASED | 3201208 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Motor Arm Left Score | Motor arm left | NINDS | caBIG | | RELEASED | 3204316 | 1.0 |
| <input type="checkbox"/> | NIH Stroke Scale Visual Score | Visual | NINDS | caBIG | | RELEASED | 3201211 | 1.0 |
| <input type="checkbox"/> | Vineland Adaptive Behavior Scales Performed Assessment Score Value | What was the vineland adaptive behavior scales value? | NHLBI | DCP,caBIG | | RELEASED | 3057303 | 1.0 |
| <input type="checkbox"/> | Vineland Adaptive Behavior Scales Performed Indicator | Were the vineland adaptive behavior scales performed? | NHLBI | DCP,caBIG | | RELEASED | 3057299 | 1.0 |
| <input type="checkbox"/> | Vineland Adaptive Behavior Scales Performed Type | What is the vineland adaptive behavior scales performance | NHLBI | DCP,caBIG | | RELEASED | 3085846 | 1.0 |

Refresh tree

- caDSR Contexts
 - ABTC (Adult Brain Tumor Consortium)
 - AECC (Albert Einstein Cancer Center)
 - Alliance (Alliance)
 - BOLD (Breast Oncology Local Disease)
 - BRIDG (BRIDG Collaboration)
 - caBIG (NCI cancer Biomedical Informatics Grid)
 - Classifications
 - 10 PROMIS Global Health Questionnaire

- PhenX
 - Anthropometrics
 - ATOS (Alcohol, Tobacco, and Other Substances)
 - Cancer
 - Cardiovascular
 - Demographics
 - Diabetes
 - Environmental Exposures

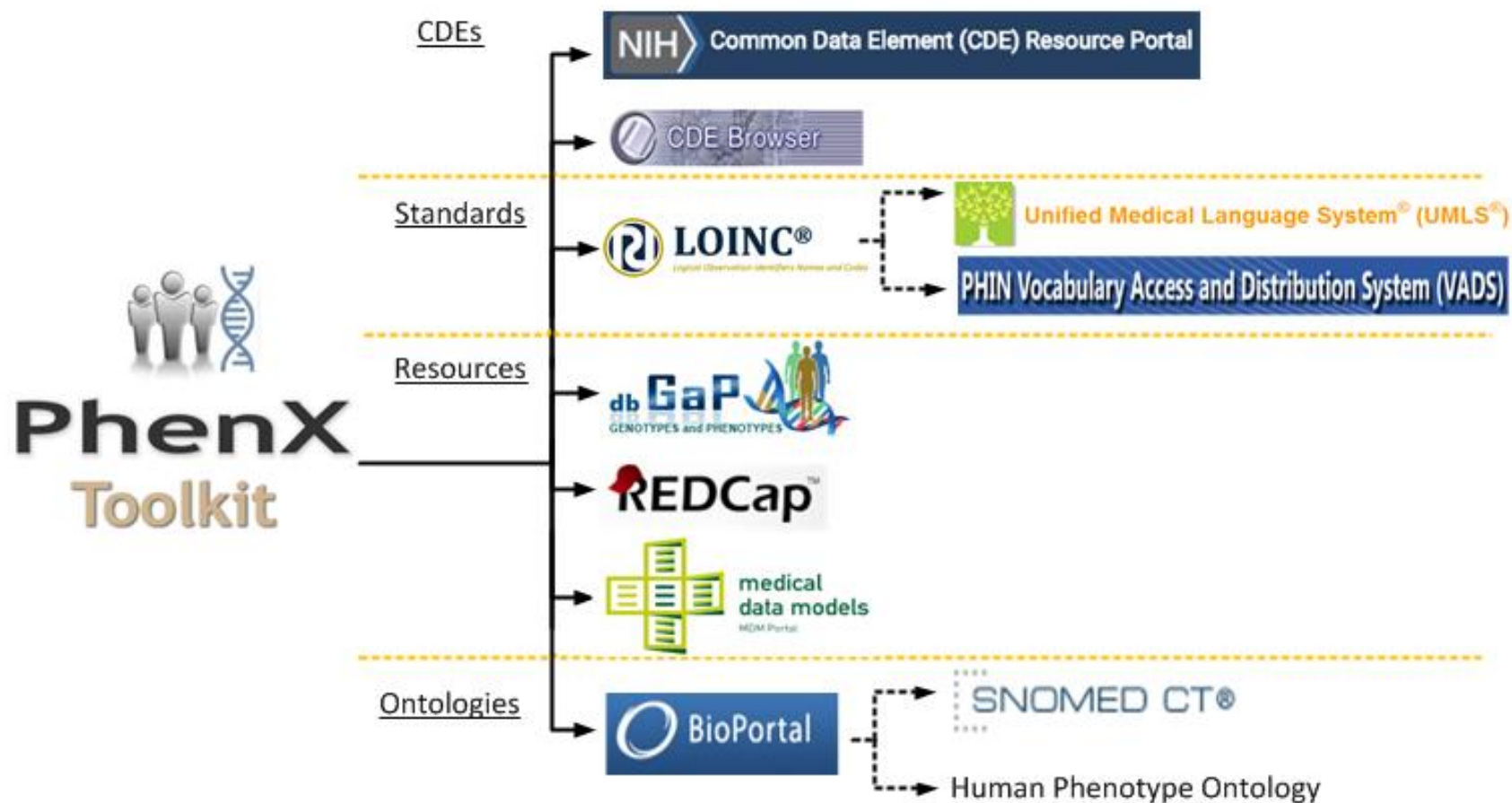
- Physical Activity and Physical Fitness
- Psychiatric
- Psychosocial
- Rare Genetic Conditions
- Reproductive Health
- Respiratory
- Sickle Cell Disease Collection
 - Sickle Cell Disease Core Tier 1:
 - Sickle Cell Disease Core Tier 2:
 - Sickle Cell Disease WG 1: Cardiovascular, Pulmonary, and Renal
 - Sickle Cell Disease WG 2: Neurology, Quality of Life, and Health S

Toward Data Interoperability: Cross-reference Table for PhenX Measures and Variables

- dbGaP, by mapping at the variable level (by PhenX domain)
- LOINC codes for the first 21 domains at the domain, measure, protocol and variable levels
- caDSR CDEs for each PhenX measure at the protocol level

| PhenX variable | PhenX ID | dbGaP variable | LOINC Code | caDSR CDE |
|----------------|----------------|----------------|------------|-----------|
| Current Age | PX010101020000 | phv00023938.v1 | 21612-7 | 2423393 |
| Ethnicity | PX010501010000 | phv00023941.v1 | 56050-8 | 2200284 |
| Gender | PX010701010000 | phv00023939.v1 | 46607-8 | 2179640 |

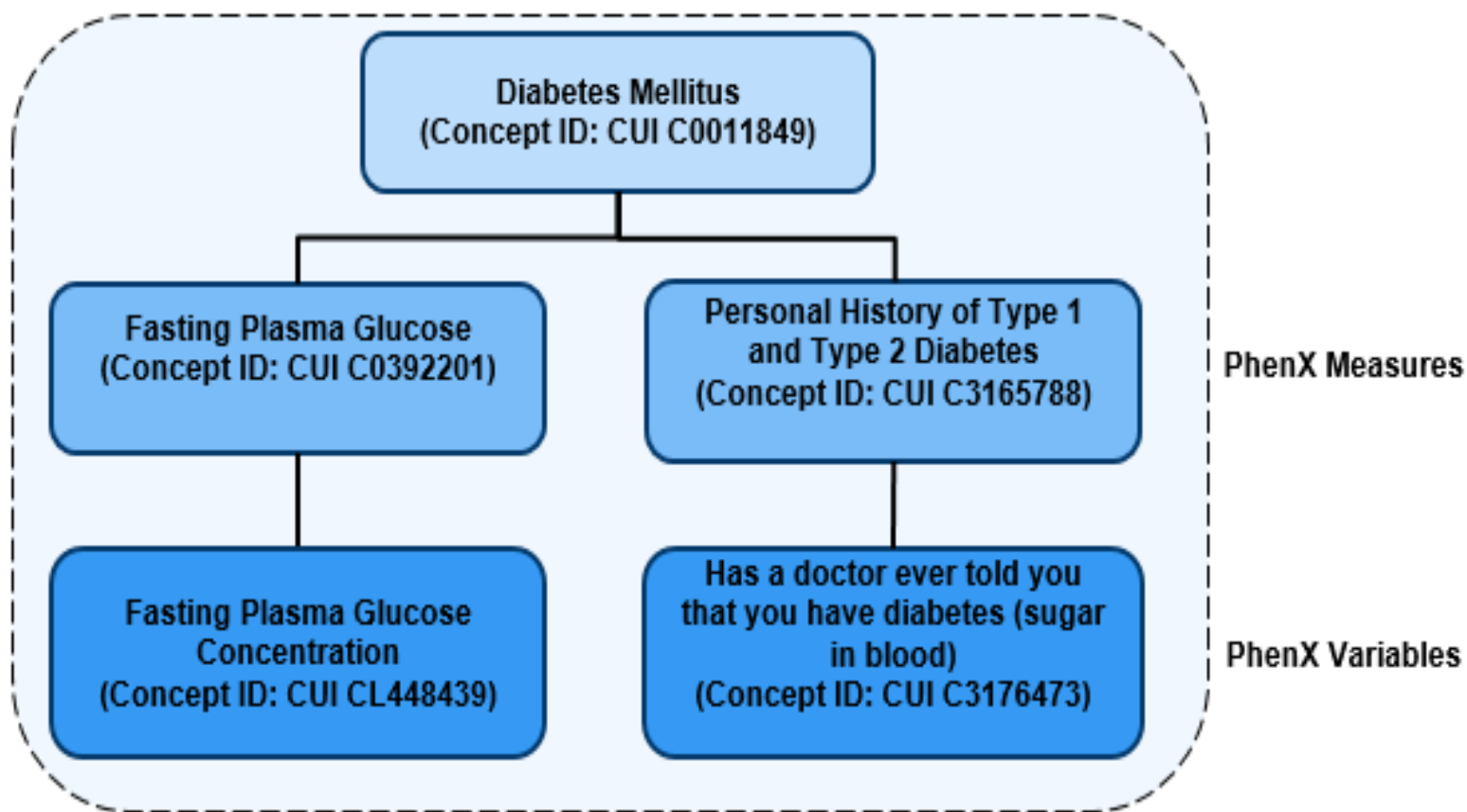
PhenX Measures are Connected to Standards and Resources



PhenX and Data Harmonization

- Use of PhenX measures reduces the need for data harmonization
- Aim is to enhance data interoperability, via standards and collaborations
- Looking to extend PhenX ontology (supports SQT) to support development of data harmonization tools

Ontology Relationships between the PhenX Variables



PhenX Toolkit in the Community

- Registered Users (2,286)
- Funding Opportunity Announcements (184)
- NIH Initiatives and Funded Studies

PhenX Measures Being Included in Current Initiatives and NIH-Funded Studies

| NIH Research Initiative/Study | Number of PhenX Measures | Sample size |
|--|--------------------------|-------------|
| Adolescent Brain Cognitive Development (ABCD) Study | 43 | 10,000* |
| Precision Medicine Initiative (PMI) | 16 | 1 million* |
| Population Assessment of Tobacco and Health (PATH) | 16 | 46,000 |
| Tobacco Centers of Regulatory Science (TCORS) | 38 | unknown |
| Environmental influences on Child Health Outcomes: Patient Reported Outcomes Research Resource Center Core (ECHO PRO Core) | PhenX recommended in RFA | 50,000* |
| Data Coordinating Center for Sickle Cell Disease Implementation Consortium (SCDIC): | PhenX recommended in RFA | 2,100* |
| * Planned | | |

Acknowledgements

- NHGRI
 - Erin Ramos (Project Scientist)
 - Margaret Ginoza (Program Analyst)
- PhenX Steering Committee
 - Mary Marazita, Co-Chair
 - Cathy McCarty, Co-Chair
- WG Chairs / Members
- NIH Liaisons
- Other Liaisons, HHS, DoD, CDC, FDA, VA
- NIDA lead – Kevin Conway
- TRSP lead – Kay Wanke
- NIMH lead – Greg Farber
- NHLBI lead – Ellen Werner
- RTI Team
 - Carol Hamilton (Principal Investigator)
 - Tabitha Hendershot (Co-Investigator)
 - Amanda Riley (Project Manager)
 - Darigg Brown
 - Wayne Huggins
 - Debbie Maiese
 - Destiney Nettles
 - Helen Pan
 - Mike Phillips
 - Toolkit team
 - Communications team
 - Logistics team
 - Collaborators

Resources

- www.phenxtoolkit.org
 - Find PhenX measures for inclusion in your study
 - Quick Start guide
 - Tutorial
- 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

Questions?

