



PhenX Toolkit

consensus measures for Phenotypes and eXposures

<https://www.phenxtoolkit.org>

Rare Genetic Conditions Measures for PhenX Toolkit

	Measure	Description of Measurement Protocol
1	Ataxia Rating Scale	The Scale for the Assessment and Rating of Ataxia (SARA) is a clinical scale used to assess cerebellar ataxia. The scale includes eight items that are related to gait, stance, sitting, speech, finger-chase test, nose-finger test, fast alternating movements, and heel-shin test.
2	Body Proportions	Arm span-to-height comparison is determined by measuring the individual's arm span and standing height and then comparing the two measurements (e.g., arm span is 3 centimeters greater than height).
3	Bone Age	The latest Tanner and Whitehouse Method, sometimes referred to as TW2, involves the interpretation of a radiograph of the left hand and wrist by a trained radiologist.
4	Child Oral Health Pain	The Child Oral Health Impact Profile–Short Form 19 (COHIP-SF 19) is a 19-item, interviewer-administered questionnaire about the oral health of a child to assess the child's quality of life.
5	Complete Blood Count (CBC)	The Complete Blood Count (CBC) protocol is a standard blood panel from the National Health and Nutrition Examination Survey (NHANES) and is performed on participants ages 1 and older. Blood is collected from participants via standard venipuncture, and an ambient blood sample is placed into a hematological analyzer for CBC and differential analyses.
6	Disability Index	The Oswestry Disability Index (ODI ©) version 2.1a is a 10-item, proprietary, self-administered questionnaire that asks questions regarding how an individual's back (or leg) trouble affects the ability to perform routine daily activities.
7	Disease Progression and Regression	The Newcastle Paediatric Mitochondrial Disease Scale (NPMDS) and the Newcastle Mitochondrial Disease Adult Scale (NMDAS) can be used to evaluate the progression of mitochondrial disease. There are three versions of the NPMDS, each for a specific age range (0–24 months, 2–11 years, and 12–18 years). The NMDAS is for adult patients over 16 years.
8	Disorders of Respiratory Control with Inherent Autonomic Dysregulation	The American Thoracic Society (ATS) policy statement regarding Congenital Central Hypoventilation Syndrome (CCHS) is a comprehensive guide for diagnosing an individual with CCHS by evaluating his or her phenotype and determining their paired-like homeobox gene (PHOX2B) genotype.
9	Echocardiography Phenotypes	The British Society of Echocardiography (BSE) Education Committee guidelines for a standard transthoracic echocardiogram (TTE) consist of a minimum data set and a recommended sequence on how to perform a comprehensive assessment.
10	Family Health History	My Family Health Portrait (MFHP) is a free, Internet-based tool that allows individuals to record their family health history (in the form of a pedigree) and that is used to collect information regarding the individual and his or her close biological relatives.
11	Growth Charts	The World Health Organization (WHO) and Centers for Disease Control and Prevention (CDC) growth charts include standard graph lines and a series of percentile curves that demonstrate the distribution of certain body measurements.
12	Quality of Life	<p>Pediatric: PROMIS© Version 1.1 Pediatric Profile 25 consists of seven item banks: physical function mobility, anxiety, depressive symptoms, fatigue, peer relationships, pain interference, and pain intensity.</p> <p>Adult: This protocol includes 29 self-administered, quality-of-life-type questions from the PROMIS® Profile 29 for adults. The quality-of-life questions include physical function, anxiety, depression, fatigue, sleep disturbance, ability to participate in social roles and activities, and pain intensity.</p>
13	Scale of Developmental Domains of Early Childhood	The Mullen Scales of Early Learning includes five scales that provide information on an infant's or child's cognitive and motor ability and also assess a child's readiness for school.
14	Scoliosis - Physical Assessment	The aggregate protocol includes the Adam's forward bend test and an x-ray with Cobb technique to determine if an individual has scoliosis.
15	Scoliosis - Quality of Life	The Pediatric Outcomes Data Collection Instrument (PODCI) consists of 86 items and is designed to collect data regarding an individual's general health and problems related to bone and muscle conditions.
16	Sweat Chloride Test	Quantitative pilocarpine iontophoresis is the procedure also known as the sweat chloride test. It involves stimulation of sweat, collection of sweat onto gauze or filter paper, measurement of the weight or volume of sweat collected, and analyses to determine the sweat chloride concentration.

NOTE: Complete protocols and links to common data elements are available through the PhenX Toolkit at <https://www.phenxtoolkit.org>.

What is the PhenX Toolkit?

The PhenX Toolkit is an online catalog of recommended, standard measures and is available for use at no cost at <https://www.phenxtoolkit.org>.

The purpose of the PhenX Toolkit is to:

- Provide recommended, standard measures of phenotypes and exposures for use in biomedical research
- Facilitate acceptance and use of standard PhenX measures
- Promote collaboration and facilitate cross-study analyses

PhenX Toolkit features:

- The Smart Query Tool provides two search options: a Smart Search based on keywords (and synonyms) and a Text Search that searches all text
- Browse options: Domains, Measures, Collections, Supplemental Information, and hierarchical tree view
- Register your study to find other researchers using the same measures and to explore opportunities for cross-study analysis
- Standards included: cancer Data Standards Registry and Repository (caDSR) Common Data Elements (CDEs) and Logical Observation Identifiers Names and Codes (LOINC)
- Registered Users have access to additional features and functionality, e.g., saving more than one “My Toolkit”
- Quick Start and Tutorial resources help new users become familiar with the Toolkit
- Annotation Tool in Toolkit Resources: Annotate the PhenX protocols that you use to assess rare genetic diseases and conditions
- REDCap Instrument Zip files for PhenX protocols can be uploaded directly to REDCap



PhenX Register Your Study

The PhenX Register Your Study feature makes it possible for Toolkit Registered Users to see what PhenX measures other investigators are using. After you register and log in, you can register your study by filling out a short form with the following information:

- Name of funded study
- Selected PhenX protocols
- Principal investigator
- Primary contact name
- Primary contact email
- Number of participants
- Funding source
- Primary research focus

This will only take you a few minutes and will foster collaboration and cross-study analysis.

If you need assistance, please email contact@phenxtoolkit.org, and we will be happy to help you Register Your Study.



PhenX Research Domains

- Alcohol, Tobacco, and Other Substances
- Anthropometrics
- Cancer
- Cardiovascular
- Demographics
- Diabetes
- Environmental Exposures
- Gastrointestinal
- Infectious Diseases and Immunity
- Neurology
- Nutrition and Dietary Supplements
- Obesity
- Ocular
- Oral Health
- Physical Activity and Physical Fitness
- Psychiatric
- Psychosocial
- Rare Genetic Conditions
- Reproductive Health
- Respiratory
- Skin, Bone, Muscle, and Joint
- Social Environments¹
- Speech and Hearing

¹ Funding for the Social Environments domain provided by the Office of Behavioral and Social Sciences Research (OBSSR).



Annotate PhenX Measures

PhenX developed a web-based Annotation Tool to collect information about the use of PhenX measures to assess rare genetic diseases and conditions.

If you would like to enhance the PhenX measures by providing information about rare genetic conditions or other diseases, please use the Annotation Tool under the “Resource” navigation bar. There are five steps to using the Annotation Tool:

1. Select a condition
2. Select a PhenX measure by browsing or searching
3. Provide a publication citation
4. Add comments
5. Hit the “Submit” button

By annotating PhenX measures, you will help the biomedical community use PhenX measures to assess rare genetic conditions and promote using of common measures.

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Dr. Carol M. Hamilton is the RTI International Principal Investigator, and Dr. Erin M. Ramos is the NHGRI Project Scientist.

More information is available at the project web portal: <https://www.phenx.org>