**Introductory Call #2 to the PhenX Genomic Medicine Implementation Working Group (WG)**

**Meeting Minutes**

October 11, 2019

**Attendees**

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| **WG Members**Kyle Brothers, Co-chairAngela BradburyLori Orlando**SC Liaison**Marylyn Ritchie | **NHGRI**Natalie PinoKerry Goetz | **RTI Staff**Tabitha HendershotDebbie MaieseMichael PhillipsJennifer SchodenLynda Grahill |
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**Action Items and Decisions**

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| **Action**  | **Responsible Person** |
| Send the WG members the revised scope and assignments for the in-person meeting | Jennifer Schoden |
| Prepare a list of relevant genomic medicine websites and consortia. | Mike Phillips |
| Send Mike Phillips suggestions for genomic medicine websites. | Erin Ramos, NIH |
| Return Contact Form, Bio Sketch, and Travel Form to RTI by October 18 for the November 18 in-person meeting.  | WG members |

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| **Decisions**  |
| Add scope element for Implementation Science: Baseline and Follow-Up. WG member assignments to scope elements: *Education (Patient and Provider)*: Dr. Zierhut*Change in Management and Treatment*: Dr. Chanprasert*ELSI (Ethical Legal Social Implications Genome Projects)*: Dr. Brothers*Return of Results*: Dr. Chung*Patient Outcomes*: Dr. Torkamani*Impact of Intervention on Cost and Utilization of Healthcare*: Dr. Ritchie noted the importance of this element and the SC may consider as a new domain *Implementation: Baseline and Follow-Up*: Drs. Orlando and Bradbury |

**Welcome and Introductions**

Dr. Kyle Brothers Working Group (WG) co-chair) gave an introduction, noting this is the second introductory call. He asked the meeting attendees to introduce themselves.

Dr. Brothers is a general pediatrician and bioethicist at the University of Louisville. He has done work in electronic medical records and genomics (eMERGE), Clinical Sequencing Evidence-Generating Research (CSER), and is involved in translation of genomics into clinical settings extended into real life outcomes. He has also done work on research ethics related to DNA biorepositories.

Dr. Angela Bradbury is an Assistant Professor of Medicine at the Hospital of the University of Pennsylvania and, by training, a medical oncologist and cancer genetics researcher focused on the implementation of genetics research.

Dr. Lori Orlando is an internist and an Associate Professor of Medicine and Director of the Precision Medicine Program in the Center for Applied Genomics and Precision Medicine at Duke University. Her work focuses on the use of family health history to identify hereditary syndromes and assess risk. She has worked through IGNITE (Implementing Genomics in Clinical Practice) and to push implementation science through the National Human Genome Research Institute (NHGRI) to help practitioners understand why more implementation science is needed.

Dr. Marylyn Ritchie, who sits on the Steering Committee (SC) of PhenX with Dr. Erin Ramos, is a professor of genetics at the University of Pennsylvania. Her research links health records with genomics. She emphasized the importance of the work of PhenX, noting the value of harnessing available data.

Ms. Natalie Pino is a program analyst at NHGRI working with Dr. Erin Ramos.

Dr. Kerry Goetz is a liaison to PhenX from the National Eye Institute (NEI). She follows PhenX for updates on inherited eye diseases and for social determinants in eye disorders.

Ms. Tabitha Hendershot is the Co-Investigator of PhenX.

Ms. Debbie Maiese is the PhenX Consensus Coordinator and has worked for PhenX since its inception.

Mr. Michael Phillips is the WG Supervisor and he, along with Jennifer Schoden, will interact the most with WG members. Mr. Phillips noted that Ms. Lynda Grahill is serving as the notetaker for the WG meetings.

Ms. Jennifer Schoden is the WG Manager. She has worked with PhenX for about a year and a half.

**Overview of PhenX**

Ms. Hendershot reviewed the PowerPoint overview of PhenX, noting that NHGRI has funded RTI’s work on PhenX since 2007 through two grants. The idea behind PhenX is NHGRI’s need to have standard measures and protocols for genome-wide association studies (GWAS). Standard measures facilitate cross-study analysis. The PhenX Toolkit is a catalog of recommended measurement protocols. Toolkit content is developed through the WGs, which reach consensus after discussion. Toolkit content is available free; but some protocols may be proprietary. Ms. Hendershot noted that the home page of the Toolkit will be refreshed in the next week or so - its appearance may differ from that in the PowerPoint. The Toolkit has 25 domains or topics, with almost 800 data collections protocols spread across those domains. The SC provides guidance to the WGs and chooses the domains. Representatives from National Institutes of Health’s (NIH) Institutes and Centers act as liaisons or can become members of a WG. Expert Review Panels (ERPs) are subject matter experts who review the content of domains to ensure that it remains scientifically valid and includes the best.

A domain is a larger topical area with a unifying theme. A protocol is how you measure the data to be collected, or what the WG is asked to provide. The protocol is the end result of this WG process, or what ends up in the Toolkit—a validated survey measure, an instrument, or any material about how that measure is implemented is captured as a protocol of the Toolkit.

**Guidance Document for Working Groups**

Mr. Phillips presented the guidance document for WGs, which has all the detail on definitions and WG processes, including details on the data sheets that the WG will be developing. The WG will have to identify the scope elements for the Toolkit and consider related measures already included, such as quality of life measures. As the WG considers the scope, members should review identified gaps in the domain and its measurement protocols and recommendations for protocols. Selected protocols should be clear, well established, and broad; some protocols come from major studies such as National Health and Nutrition Examination Survey (NHANES) and ECHO (Environmental Influences on Child Health Outcomes), or other standards (publicized). The WG should consider the burden on both participants and investigators and determine the protocol’s application—is it for adults or children? Men or women? The WG can recommend multiple protocols for a scope element. Data sheets have a burden table that helps define high-burden protocols (e.g. special equipment, training, collection requirements).

At the in-person meeting, WG members will make a presentation of their scope element and recommend protocols. Presentations will be about 15 minutes, with 5–10 minutes for discussion. WG members should review existing protocols in the PhenX Toolkit to determine whether a particular scope element is already covered. For outreach, the WG will narrow down proposed protocols to 15–18, only 4 of which can be high burden. After outreach, the WG will recommend to 15 measurement protocols 2 of which can be high burden. The WG need not propose a new measure for every scope element; new measures should not compete with existing measures.

At the in-person meeting November 18, WG members will present their protocol suggestions and the WG will make its selections for outreach. During the meeting, Ms. Schoden will document the recommended protocols on a spreadsheet to help the WG keep track and make its determination. After the in-person meeting, Ms. Schoden and Mr. Philips will prepare the data sheets for the WG’s review. The data sheets will go to Outreach January 2020. The WG will have a teleconference to discuss the outreach feedback and prepare final recommendations for the SC.

**Scope of the Genomic Medicine Implementation Domain**

Review Draft Scope

Dr. Brothers reviewed the draft scope of the Genomic Medicine Implementation domain and the scope elements: *Education, Change in Management and Treatment, ELSI, Return of Results, Patient Outcomes,* and *Impact of Intervention on Cost and Utilization of Health Care*.

Additions, Deletions, and Refinements to the Scope

Dr. Orlando felt there was a considerable amount left out of the draft scope, for example, measuring uptake of genomic medicine interventions by providers and by patients. Dr. Brothers thought this might be covered by *Impact of Intervention.* Dr. Orlando felt that this would be more of an after-intervention measure; she thought a pre-implementation assessment would be more important.

Dr. Orlando agreed to research this scope element, which would be called Pre-implementation Assessment.

With further discussion, the WG revised this decision, noting that the issues to be addressed by the element—study design, how clinical care might be changed, demographic considerations, uptake (of counseling, testing, the cascade of relatives, declination, etc.)—seemed more suited to a broader Implementation scope element.

Dr. Brothers introduced the issue of cost and utilization, broached on the previous introductory call, and how it might be accommodated in the current scope elements. Dr. Orlando felt that cost-effectiveness modeling may be standardized already. Ms. Maiese noted that the Toolkit may have some cost- measures for substance abuse research. Dr. Bradbury felt that cost would be an important consideration to include in the Toolkit. Dr. Ritchie noted that the economics of genomics may need more standardization. Dr. Brothers noted that cost could be a broad enough issue for a WG of its own. Dr. Orlando agreed and Dr. Bradbury noted that it would be important. Dr. Brothers noted that they might recommend that the SC proceed with cost as another, separate domain.

**Decision**: The final new scope element decided upon was *Implementation: Baseline and Follow-Up*.

Identify WG Member for Each Scope Element

*Implementation: Baseline and Follow-Up:* Dr. Orlando will do Baseline and Dr. Bradbury will do Follow-up.

Dr. Brothers asked if the WG might want to remove the redundancy in other scope element assignments: *Education* would be assigned to Dr. Zierhut; *ELSI*, Dr. Brothers; *Change in Management*, Dr. Chanprasert; *Return of Results*, Dr. Chung; and *Patient Outcomes*, Dr. Torkamani.

*Impact of Intervention on Cost and Utilization of Healthcare*: Dr. Ritchie noted this might be its own domain.

 **Decision:** Assignments were made for the scope elements as listed above.

**Decision:** Do not include the scope element “Impact of Intervention on Cost and Utilization of Healthcare” with this WG.

**WG Member Presentations at In-Person Meeting**

Review Presentation Template

Mr. Phillips introduced the template for a sample presentation file in the meeting materials.

Sample Presentation

Mr. Phillips reviewed the sample presentation from a member of the Pregnancy WG. WG members can include a PDF file of documentation if PowerPoint does not accommodate the protocols. RTI can assist with presentations and answer any questions. WG members may work together on presentations.

Identification of Protocol Sources

Mr. Phillips noted that Dr. Ramos had shared some consortia websites from CESR, IGNITE, etc., for WG members to use to identify protocols. He noted that measures already in the Toolkit that might apply to this domain are quality of life measures.

**Next Steps**

Goals for November 18 In-Person Meeting

Ms. Schoden reviewed the expectations for the in-person meeting on November 18 at the Hyatt Regency in Bethesda, Maryland. The WG will finalize the list of measures for outreach after discussion of each measure. Ms. Maiese noted that having nothing to recommend for a scope element is acceptable. Recommendations often change through the WG discussion or through follow-up after the in-person meeting by email or further calls, if necessary.

Contact Forms, Travel Forms, and Other Logistics

Ms. Schoden noted that WG members should return their contact sheets and biosketches, if they have not done so. Travel forms should be submitted to Stacie White at stwhite@rti.org.

**Adjourn**

The meeting was adjourned at 2:11 p.m. Eastern Daylight Time.