

PhenX Steering Committee (SC) Meeting Minutes

January 13, 2021

Attendees

SC Members

Mary Marazita, Co-chair
 Cathy McCarty, Co-chair
 Lindsay Farrer
 Elaine Faustman
 Jonathan Haines
 Tabitha Hendershot
 Erin Ramos
 Marylyn Ritchie
 Sharon Terry
 Rosalind Wright

National Institutes of Health (NIH)

Nishadi Rajapakse
 Mariela Shirley
 Phil Tonkins
 Kay Wanke

RTI Staff

Carol Hamilton
 Lisa Cox
 Lisa Gektland
 Lauren Gridley
 Wayne Huggins
 Stephen Hwang
 Michelle Krzyzanowski
 Debbie Maiese
 Mark Nelms
 Helen Pan
 Mike Phillips
 Amanda Riley
 Justin Waterfield
 Pat West
 David Williams
 Thien Lam

Guest

Ian Terry

National Human Genome Research Institute (NHGRI)

Jyoti Dayal
 Madison Goldrich

Action Items	Responsible Person
Use social media to disseminate bite-size pieces of information on PhenX	Pat West
Include the nine-question Coronavirus Racial Bias Scale in the COVID-19 NAME collection	Michelle Krzyzanowski
Find out more about DR2 meeting that occurs every 3 years (Faustman, NIEHS)	Carol Hamilton
Invite Catherine Gordon to be member of the Bone & Joint WG	Tabitha Hendershot
Review COVID-19 modules to check for any others with aggregate scoring (should not be broken up)	Michelle Krzyzanowski
Decisions	
Bone & Joint WG will be allocated 15 measures in total	
ERPs to be renamed "Content Expert Panels"	

I. Welcome and Goals of the Meeting

Mary Marazita started the meeting at 10:04am Eastern Time. This is the first of a two-part meeting this January.

II. NHGRI Update

Erin Ramos welcomed everyone on behalf of NHGRI and the PhenX team. She thanked everyone for their dedication to PhenX, noting that some SC members have served for 10 years. Her update mostly focused on the NHGRI strategic vision, published October 2020. She recommended people look at the NHGRI COVID-19 web page, which has a link to the PhenX Toolkit in the resources highlights page.

Erin moved on to the NHGRI strategic plan, covering the history and context, planning process, and an overview of the highlights. There have been three strategic plans between 1990 and 2020. The first NHGRI plan started with the Human Genome Project.

Acknowledging that she was using Dr. Eric Green's slides, Erin compared NHGRI funding with overall NIH funding for human genetics. NHGRI is one of the smaller institutes in NIH, though funding increased 10-fold since 1990. Initially, NHGRI funded 95% of NIH genetics research in 2020, NHGRI is only funding 10%. This is exciting because the genomic work that began with NHGRI is now occurring across the NIH. NHGRI is now considering how to push the field and think about what is at the forefront that would be beneficial to the rest of NIH.

NHGRI started the latest round of strategic planning in 2018. NHGRI held workshops, and town halls to gather feedback on the strategic vision. The plan was published in *Nature* (<https://doi.org/10.1038/s41586-020-2817-4>) to coincide with the 20th anniversary of the Human Genome Project. It emphasizes (1) guiding principles and values for human genomics, (2) sustaining and improving a robust foundation for genomics research, (3) breaking down barriers that impede progress in genomics, and (4) compelling genomics research projects in biomedicine. The strategic plan closes with bold predictions for human genomics by 2030. Erin highlighted that NHGRI has a new seminar series for each bold prediction. The first is on February 1, 2021 3-4:30pm.

NHGRI just published an action agenda for building a diverse genomics workforce. The goals are to (1) develop and support initiatives that provide early exposure and access to careers in genomics; (2) develop and support training programs and networks that connect undergraduate education to careers in genomics; (3) develop and support training, career development, and research transition programs that lead to independent research and clinical careers in genomics; and (4) evaluate progress towards achieving greater diversity in the genomics workforce.

Elaine Faustman said the comments about where genomics funding lies across NIH is fascinating. It bolsters the comments Erin made about genetics research in general. Debbie Maiese said Dr. Eric Green and Vence Bonham recently published a commentary on the NHGRI workforce diversity action agenda. (<https://doi.org/10.1016/j.ajhg.2020.12.013>

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III. RTI Update

Carol Hamilton explained the RTI update would be about PhenX Toolkit releases, the status of working groups (WGs) and expert review panels (ERPs), manuscripts in progress, social media presence—getting SC's ideas about what has worked—and setting the stage for the January 29 meeting.

New Toolkit releases included COVID-19 collections. People worked hard to get it out in October. Michelle Krzyzanowski will talk about it later. The cancer outcomes and survivorship protocols were also released.

Sickle Cell Disease has two WGs in progress. The Curative Therapies WG is having a meeting this Friday. The Pain WG will soon have a meeting to review the scope.

Regarding publications, the Genomic Medicine Implementation WG submitted a manuscript were to *Nature, Genetics in Medicine*. A manuscript about the COVID-19 collections was submitted to *Current Protocols in Human Genetics* on December 20, 2020. Speech, Language, and Hearing WG

submitted a manuscript to *Annals of Human Genetics*. The draft Social Determinants of Health went out to WG members to get their input. The manuscript will be submitted to *JAMA*. Nishadi Rajapakse said the commentary has been with her Institute Director for a few months. She hopes he will provide feedback soon. Carol said a manuscript for pediatric development is almost ready to submit to *Pediatrics*. A manuscript is being developed for *Current Protocols in Human Genetics* that will be an update to the PhenX Toolkit manuscript that was published in 2015. The database of Genotypes and Phenotypes (dbGaP) manuscript is nearly ready for submission to the *Journal of the American Medical Informatics Association*.

Tabitha Hendershot presented on PhenX's social media presence. Feedback is requested. The PhenX Twitter handle is @PhenXToolkit and has 41 followers. The Twitter account really started seeing use on May 12, 2020, with the Social Determinants of Health release. There is a PhenX Toolkit channel on YouTube with 12 subscribers. There is a question of how to encourage people to follow PhenX on Twitter and YouTube and make it easy for them to do so. Is there other content to offer?

Tabitha said she was trying to pick SC members' brains about approaches to increase visibility on social media. One item being considered is to make it more clear on the website to follow PhenX on Twitter and YouTube. Additionally, that information could be included in the newsletter so people can access it or follow and subscribe from the newsletter when they read it. Are there people on this call that might be willing to retweet content? Are there groups PhenX might follow to improve online relationships? What else might PhenX do beyond what it is doing?

Marylyn Ritchie said she didn't know that we had Twitter. She noticed that the PhenX Toolkit only follows three Twitter accounts. One gains followers by following others. A lot of journals, NIH institutes, universities, and scientists are on Twitter. Marylyn herself does not tweet a ton but is happy to retweet if she is sent an email saying PhenX has an important tweet and asking people to retweet, she would be happy to do that. Tabitha asked if that would be helpful for others. Sharon Terry said that Genetic Alliance has 20,000 followers. She finds Twitter not so useful. Facebook, Twitter, and WhatsApp will decrease in popularity. Millennials are not using these platforms anymore. But she is happy to retweet.

Carol inquired where the millennials were going. Sharon replied that TikTok is huge, but that is not where PhenX wants to advertise. People are not looking to those places for work things; it is more of a social platform. Some people are using LinkedIn. But there is no easy silver bullet. Marylyn mentioned she hardly used LinkedIn but is now constantly getting inundated. Sharon noted that Ian Terry said LinkedIn is huge for his generation. —Elaine added that at the school of public health, they use ResearchGate and LinkedIn a lot. She noticed PhenX does not have blogs. There are a ton of health-related blogs.

Sharon wondered about Reddit. Ian responded that Reddit has a very interesting community in that it is borderline conspiracy theorists, but the Reddit community killed its own QAnon channel without Reddit doing anything about it. Getting Redditors to get into a subreddit can be difficult. Instagram is on the downstream but is still strong in comparison to Facebook and Twitter. Snapchat is on its way out and never really had the professional presence other platforms had. Some publications are coming out about how much people engage with professional content on TikTok.

Erin said if PhenX does use Twitter, to come up with ways to disseminate bite-size pieces of information. She agreed with following people to make connections. Carol noted that PhenX will see what it can do. It would be nice to have a social media presence which it does not have now.

Carol continued with a heads up for what is coming down the pike. PhenX is doing a needs assessment, getting ready for the renewal proposal due May 2021, and doing a PhenX citation analysis. There will be a two-part launch of the sex and gender minorities project. Karen Parker may be asked to come talk to SC. PhenX will also present on the Polygenic Risk Score guidance being proposed by the Smoking Cessation WG. RTI is proposing a workshop for ASHG. PhenX will highlight

the NHGRI strategic plan in the renewal proposal. The PhenX portal is being updated. PhenX is working on a better way to do outreach. Two of the Sickle Cell Disease WGs are well underway; there will be four WGs going at the same time. The thing that really matters is the people who work on PhenX. They actually care. They want it to make a difference. You cannot make people care but when they do it shows.

IV. COVID-19

Michelle provided updates on the COVID-19 supplement. Six COVID-19 specialty collections were released October 30, 2020: Behaviors and Risks; Ethnicity, Race, and Demographics; History, Treatment, and Outcomes; Information Resources; Psychosocial and Mental Health; and Socioeconomic. There is a button on the home page similar to other collections. The COVID-19 page was renamed to the COVID-19 Protocol Library and key word search functionality was added. The PhenX team assigned keywords to all PDF files. For example, a search on fever brings up modules and protocols related to fever. The top 10 PhenX protocols and domains for October 1-December 31, 2020 were reported. They 10 all in the Social Determinants of Health: Core collection.

A module from the Coronavirus Racial Bias Scale was included in the COVID-19 collection. The current protocol only uses two of nine questions. The original authors and feedback from NIMHD and NIH suggested expanding to use the full 9-item scale. Carol asked whether there were any objections to including the full nine questions. There were no concerns. Carol said another request from NIH was to take a look at all the COVID-19 modules to make sure things were not broken up that should not have been broken up.

Michelle stated that a COVID-19-related manuscript was submitted to Current Protocols in Human Genetics on December 20.

The goal is to be able to compare similarities across COVID-19-related questionnaires or surveys. Carol mentioned that when COVID-19 protocols were submitted to NIH, Bill Riley's team carved out modules, which went to PhenX and Disaster Research Response (DR2). They are an important part of how this effort developed. Elaine brought up an opportunity to make sure PhenX is at a meeting being planned by DR2. It is a meeting that takes place every three years for groups that work in the field. Carol said that was very helpful. PhenX has not had much contact with DR2 but will touch base. Elaine noted that she is on the planning team and has monthly calls. DR2 got an NIH conference grant.

V. ERP Updates

Tabitha gave an update on the Cancer ERP. When SC members last met, they talked about the need to review the Cancer domain and were interested in doing something with a crowdsourcing component, building on the experience PhenX had with COVID-19 crowdsourcing. There is an opportunity to review not just the protocols in the Cancer domain but the two other Cancer protocols in the Toolkit. It is a nice opportunity to do a cross-Toolkit protocol update to cancer information.

The first step will be to identify and recruit three to four experts. This small group will help guide the process almost like a steering committee. The vision is that this group will consist of people who have been involved with PhenX before, so they already understand PhenX. The idea is to try to include a former WG member and NIH liaison. The next step is for the small group to triage protocols—to update or leave as is. Their input will be sought electronically so that input obtained without the influence of others. Then the small group would look at results and make final decisions about what needs to be updated. The small group would also be asked to identify the crowd, engaging people who are really engaged with cancer research so they can inform how content is being updated. The next step is to do crowd sourcing with the identified cancer research

community. The crowd would review only the protocols that the small group identified as needing an update. The crowd would be asked, if you do not like what is in the Toolkit now, what do you think about these options? The crowd would select from options (e.g., NHANES, ECHO protocols) of already validated instruments that are available or write in a new response. The small group will review crowd feedback and decide which protocols to recommend. RTI will develop datasheets that will go to the SC for review and approval. Then, PhenX will release updates in the Toolkit. Finally, the crowd will be informed of final decisions to see how their input was implemented in the Toolkit.

A new name is needed for the ERP because the process being implemented is different. There is a small group but there is also use of a crowd. There is an intentional effort to stay away from the word “domain” because it is PhenX jargon. The word “review” is also to be avoided as it is not compelling or interesting. Some options are Content Expert Panel, Content Review Panel, Content Update Panel, and Content Renovation Team. SC members can throw out other ideas for names or say if they think ERP is still suitable. Carol wondered whether “crowd” could be put in the name. Mary thought that the crowd is a tool that can be used to contribute to the process. There are a couple of votes in the Zoom chat box for Content Expert Panel. Content Update Panel may not be suitable because the content may or may not need to be updated. Same with Content Renovation Team; not necessarily a need for renovation. There is consensus from the SC on Content Expert Panel.

Decision: New review groups to be named “Content Expert Panels”

VI. Bone and Joint WG Update

Lisa Cox gave an update on the Bone and Joint WG. She met with Lindsay Farrer, the SC liaison for that WG, in late December. They talked about scope and potential WG candidates. Tabitha sent an exploring your interest communication to Dr. David Felson at Boston University and hopes to hear from him soon. Lindsay noted that David is working on a grant application, so he is busy, but not to give up on him. Lisa said that the next steps are to review and finalize the scope today and identify and invite potential WG members.

There were five existing protocols related to Bone and Joint and there were nine other protocols relevant to Bone and Joint in other domains. The WG would be asked to review existing Bone and Joint measures which are all older, then see what is missing. Lisa started to do that with Lindsay on a recent call. Inflammatory arthritis, clinical symptoms, rheumatoid arthritis, function related to ankylosing spondylitis, and biomarkers were identified as gaps. Rheumatoid arthritis and function related to ankylosing spondylitis were in the original scope but were not addressed by the Skin, Bone, Muscle, and Joint WG. Lindsay stated that one of the realities is that the people in the WG would be relied on to some extent to think about what scope elements are missing. They would know more. Once the WG is formed, it is expected that they will have suggestions. Lisa added that it is a quandary: needing enough of a scope to recruit the right people and recruiting the right people with the expertise for addressing the scope elements.

Carol asked in the Zoom chat box how many measures should be allocated. Jonathan Haines said the original WG was divided so this WG should get half as many, or it is a new group so it should have the full 15? He suggested letting them have the full 15. But an argument can be made in either direction. Lindsay noted that some measures already exist. What number of bone and joint compared with skin and muscle? Jonathan suggested to take the measures that were already there and let the WG add up to 15. Give the WG the flexibility to bring more measures into the Toolkit.

Elaine noticed that bone age is on the list but wondered whether the WG should investigate if there are any earlier life or developmental changes in bone and joint evaluation. It seemed to her that there might be. There is calcium intake, but that is such a basic one. Are there things that should be looked at from a developmental standpoint in terms of bone and joint? Tabitha

responded that this is focused a lot on older people, but perhaps content can be added for children. Carol noted that the guidance has always been to look at all life stages. But if a pediatrician is not on a panel, protocols for children may not be identified. Debbie inquired whether a pediatrician working on bone should be recruited. Lindsay thought that, in terms of phenotypes that are detected and assessed in younger folks, a lot of them tend to be rare. Elaine remembered recent outbreaks of undetermined origin in California. Thought to be polio, children did not go to school for fear it was contagious. She suggested looking at the Centers for Disease Control and Preventions infectious diseases involving bone and joint. Rosalind Wright also recollected what Elaine was talking about. Mary said there were some tentative WG candidates on the next slide in the presentation. Rosalind thought Catherine Gordon would be good. Approach the candidates and they usually point to others.

Debbie asked whether there were researchers to tap from ECHO. Rosalind said ECHO has a focus on growth and obesity, and not bone and joint. Debbie suggested that Lindsay put in a good word with bone and joint WG candidate David Felson. Mary said it sounded like giving the WG 15 measures had a reasonable consensus. Marylyn agreed. Mary thought it made sense since the WG was broken in half because it was too much for one group. The scope looks okay.

Decision: Bone & Joint WG will be allocated 15 measures in total.

VII. Wrap-Up

Mary stated that SC made it through the agenda for part 1. Part 2 is on January 29. The final agenda for that meeting will be distributed closer to the day.

VIII. Adjourn

The meeting was adjourned at 11:33am Eastern Time.