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all right. I think we can get started. I am delighted to welcome you to the second Lc.

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Conversations on population descriptors in clinical genetics.

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This series is in collaboration with the Ancestry Diversity working Group of Clinton, and this is the second of 3 sessions.

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My name is Sandra Sujin Lee, and I Co-direct, the Sera, the center for Lc resources and Analysis and Lc. Hub.

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With my copi. Mildred Cho. We wanted to let you know about this Lc.

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Conversations, , series as as one that's flexible in the sense that it aims to foster and engagement and open dialogue about the latest developments in Lc.

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This includes emerging research policy issues, methodologies, and other topics that are suggested to us by our growing Lc.

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Community. and so we we love receiving proposals. so please do reach out with your ideas.

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Just a few housekeeping notes. This is an hour long event we welcome questions and comments.

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Please liberally use the chat feature as a way of interacting with the panelists as well as each other.

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Also you can use the hand. raise feature to enter to ask questions.

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But when you do raise your hand and your acknowledge, if you could please lower your hand after being called on, that would be terrific.

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We really encourage for those who can to turn on your video. it's always wonderful to see each other

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One note is that we will be recording this session.

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And after the series, the the recording will be uploaded to Lc.

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Hub dot org, and it's available to the public if you have any questions.

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Please send a direct direct message to Dounia Alami Nasif, or email, as said info at Lc.

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Hub dot org Alright, so It's my pleasure to turn it over now to Alice Poke Joy, who will be introducing the series.

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Alice. Thank you so much. Sandra. It's really nice to be here for the second special of this series. I'm so grateful to Elsie Hubb to Sandra and Mildred and the Staff Rachel and Jr as well for making all of this

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possible, because these are really important conversations. So the title of this series of Lc.

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Conversations ethnicity and ancestry on chemical genetic, a laboratory test requisition for, and this is something that has been of great interest to

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The Clinton Ancestry and Diversity working group, which started in the fall of 2,017.

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As you can see, there are a lot of other things that Clinton does, and different groups that I are Involved ancestry and diversity. working group specifically is made up of scholars.

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From all different disciplines that this issue can

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Population and statistics in social science as Well, and history. probably missed a few in there, because it is so interdisciplinary. And we've been having these conversations.

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But we really wanted to. broaden the reach of what we've been talking about with regard to the use of racism and ancestry and Chemical So that's why we partnered with Elsie Hub to

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create this Lc. Conversation series. So the 3 motivating questions that we've been thinking about within the working group, and that really motivated this series. One how is information on the tester?

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Position practice. What is the most important information for clinical justice professionals to do their jobs?

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Well, all is necessary. One Is it protocol versus extraneous?

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When is it potentially harmful, and one is not collecting harmful?

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And finally, if changes to these forms were to be recommended, what are the considerations that need that we, in order to implement changes?

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What are the barriers and opportunities? So we designed A. diversity working group and partnership with Lc. Hub.

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Designed this series with 3 sessions. The first last week, was on 20 clinical lab test reposition forms in general, and thinking about population. today, we're going to be talking about the utility of population.

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So really distilling what that relevant information is among the population.

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Descriptors that serves an important purpose; and finally, our last session on the twentieth of the month will be about those revisions to

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Clinical lab requisition forms what and so as I mentioned today.

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We are working on this topic, and i'm really looking forward to the discussion. So

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With that in mind. I would like to introduce our to Singer Moderator, Dr.

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Shimida disguise, who was the Professor at Boston University. she's a national leader in a science education.

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Really I had the pleasure of meeting shamita the the station of professors of human and medical genetics, and ever since then i've been blown away at how much has on this topic in particular and is really motivated, and

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and nationally internationally recognized for her work and genetics.

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Genomic medicine covers and inclusion and and it's just that.

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We're very fortunate to have her here in moderating stuff.

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So, Shamina, please take away and thank you so much for being here, and thank you to our speakers as well.

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And thank you so much for that incredibly kind introduction alice i'm delighted to be here with everyone to discuss this incredibly important topic.

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Today we are focusing on the utility of population descriptors in clinical genetics.

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So really this is kind of the How are we doing this?

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And why are we doing this? and what is the potential benefit or harm of our approaches?

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So to kind of kick things off. But we created a few discussion questions on a jam board.

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So I think doing is going to share the link here in the chat.

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So let me just describe to you how a jam board works if you've never seen one before.

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When the link appears. What you'll do is click through on your own computer, we will thank you, Antelica.

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We will also be sharing the screen so that everybody can see the contributions of everybody on the jam board when you link through to the jamboard. You'll see a column on the left.

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We're going to stay on the first page together. and i'll tell you when we're ready to advance to the next page.

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The column on the left will include a little square icon and you can see that square icon with the text lines that will allow you to have your own post a note, and you can add your thoughts to the question, So the first question

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Here is what benefits may be realized by using a multiple choice structure to our Ea meeting race ethnicity and ancestry.

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Demographic questions,

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Yeah, seeing some a bunch of these popping up. Thanks for moving them around so that we can see all of these.

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It does look like some of the important benefits that people are recognizing is the ability to kind of group answers.

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Have some quantitative results. pulling the data easily again.

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Kind of like grouping, collecting, and analyzing harmonizing information.

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And we're also seeing a little bit of concerns which will be actually our next question.

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It looks also like, the the multiple choice assuming multiple answers are allowed, then multi-racial or multi-.

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Ethnic multi ancestry folks would also be included.

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These are all great contributions. thanks everyone so let's advance together to the next question.

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You'll see the arrow on the top of the screen So what harms may be realized by using a multiple choice structure to rea demographic questions

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Well, these are all great responses. So flattening response is planning contextual information, forcing people answer in particular categories that are predefined may not lend itself to people with complex ancestries missed opportunities for

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self-described area, reinforcing, incorrect belief and discrete categories of humans.

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These are all so important. really. Thank you so much for including these excellent contributions.

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And, by the way, even if we advance past this and you're still thinking about it.

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Then feel free to continue adding your contributions.

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I see that at least one person is having some difficulty getting into the jamboard, because there's a lot of people viewing it.

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I don't know if there's a setting for prohibiting that but hopefully, folks will be able to get in, and, as I said, you can view it again after the fact, it'll still be there.

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Okay, let's go on to the third question what benefits may be realized by using an open ended structure rather than the multiple choice structure.

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To raa Demographic questions,

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Yeah. So it looks like a lot of the contributions here are focusing on freedom or ability to answer.

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You know, in a more unique way that may reflect one's own self identity, and gets a better understanding of the full range of identities.

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These are all outstanding all right now, let's take a look at the other side of the coin.

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If we were to use on question 4 if we were to use open appended structures, what harms may be realized in using open-ended structures to ree demographic questions

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So it seems like a lot of the concerns are around analysis, because if everybody puts in their own unique descriptor, then it's hard to make kind of population wide or user-wide kind of conclusions.

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So the binning and the omb categories are coming out that we've discussed in the last session, too.

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Yeah, these are all outstanding. So thank you so much.

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These are great ways to kickstart our thinking about how and why we're using population descriptors in clinical genetics. and to get us started, We're gonna hear some summary remarks from our guest

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speakers hopefully. you've had a chance to view their talks in advance that they will give you a little bit of an overview

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Each one of them. So that will Kickstart our conversation this morning or this afternoon, depending on what time zone you're in, and our first speaker is going to be Julia Gimburn at Mail who is from Imperial

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College of London. So, Julia, if you could share your slides and get going.

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Thank you great. thank you so much. I will share my screen.

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Now

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Great. So thank you so much, everyone, for being here, and and Alice for introducing the like.

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A lot of like giving in a context to the work i'm going to be explained today.

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So I first wanted to to explain a little bit about the motivation and the approach we took for this this analysis.

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It was in collaboration with cancer genetics, and we wanted to expand the requisition form analysis.

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And and we gathered 8, 58 international forums and 61 Us.

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Forums. The first question we tried to answer is, How often are diversity measures included in clinical requisition forms?

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And And to answer this question we calculated the percentage of clinical laboratory position firms that had a question involving race at the city and ancestry, and we determined whether there was a difference between the us and in the international

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labs. The second question we try to answer is, How are diversity measures characterized in the questions?

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As so, for example, which terms are being used to describe other.

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This variable of interest, and what is the frequency of terms used across unique requisition forms?

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And finally the last question we were we looked into. How are we?

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Raises as a cnn's history data response is coded and structured, and so we assess the types and frequency of answers solicited or including in the forms. I will i'll put it on my

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pointer. Okay. So one of the first conclusions that we got to is the race. Ethnicity and ancestry is more commonly encountered in clinical care in the United States So here.

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We can see 2 pie chairs this this first one are is refers to the United States, and we see here a part of the pipe chart that's separated from the rest, and and this this is trying to show

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that i'm sorry i'm sorry to move okay this is trying to show that 30% of the clinical forms from the United States that we analyzed didn't have our ea question, whereas the rest did in contrast for

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the rest of the clinical requisition forms of the international forms.

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It was 53% of them that were in asking the question and the rest don't wear.

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So This shows how it's it's more common in the United States to be asking these kinds of questions.

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Another observation was that most of these these clinical acquisition forms in the Us.

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Were asking these questions in the form of a multiple choice question, whereas in international forums it was more common to to ask this in the blind, textful form

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In this cloud. We can observe how they're they're recorded forms from USB.

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Laboratories. we're the only ones to use race so here in Orange we see the the Us.

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Phones represented, and we see how there's only an orange bar in race because none of the international forms were asking the question using this term, whereas most of them internationally, we're asking about

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ethnicity, and most of the us forms also asks about the city, but had more granularity in the ways that we're asking the question.

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It was also interesting to see that the USB La board she's representing race of the city and ancestry in predefined category.

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So I mentioned this in a previous slide but here we can see more more clearly how it's in the us, which is this great color?

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We're they're they're mostly represented these multiple choice, with other category or without resident international.

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For we have the blind text bill. and finally, this last part of our analysis.

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We we decided to plot a coherence matrix which is basically trying to show which population descriptors are used together in requisition forms.

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And we saw how most of the time these firms include 4 categories.

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These black African, American or African white Asian and Hispanic, Latino, or Spanish, which is interesting because it's not only telling us that these are the most popular population The scriptures that are used but

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they're also somehow considered orthogonal because these are the 4 categories that are are shown as options, and and much they're they're shown, much less so.

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The rest of them are are shown, much less The ones that we see here is sometimes that we see.

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We do see them, but they're they're not shown assumptions as often as these 4, so it it begs the question, Are these the ones that we should be?

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Using. Why have we decided these 4 are the ones we want to show.

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And should these change I think that's it for me thank you thank you so much, Julia, it's a really great Kickstarter for us.

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Now, I think next we're gonna turn it over to Carly Dawson, class of 2,022 be path genetic counseling program congrats on your gradually graduation.

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And if you could go ahead and share your slides thank you I'm just pulling up my slides Now, Alright, so thank you.

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I'm excited to be a part of this panel discussion today, and I'm looking forward to our conversations. but for a brief recap of my presentation title Diversity Measures in genomic medicine I presented

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the result of a virtual workshop organized during the Wisconsin Genetics Exchange Conference.

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The purpose of this workshop was to determine how genetic professionals are using diversity measures and encourage them to consider whether current data and approaches are appropriate.

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One of our goals included collecting specific use cases or examples for which race andnicity and ancestry are utilized within the field.

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To summarize our resolve into 3 overarching areas.

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First, we ask participants to define race administration ancestry based on group concepts that were provided.

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Race was mostly concentrated with a social identity Group and ethnicity had a high concentration with a cultural group, and then ancestry had a high concentration with a biological group as well as age

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and genetic lineage Group Second, we were able to obtain some primary use cases for which race of missing ancestry is utilized.

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Within the field. he's included using the information for democrats on test requisition forms the participants also shared how labs are inconsistent and asking for this information. Other use cases included utilizing the nccn councillor

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guidelines defining carrier risks or residual risks, guiding test offerings, interpreting results insurance purposes or billing codes.

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Pedigree, risk analysis, tracking health, equity, and interpreting ultrasound findings or dismorcology.

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Third participants provided use cases for which the utility of race of missing ancestry is appropriate or inappropriate for appropriate use cases they described.

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Very interpretation providing carrier risks, test offerings, as well as under barriers or tailoring appointments for inappropriate cases for which raise the pristine ancestry are used also included

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test offerings, and whether data is self reported. participants also shared.

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How asking of this information may lead to some cycle. Social impacts and participants had also shared that using race as proxy is inappropriate.

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Since a large fraction of the Us. population is from a mixed ancestry.

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In conclusion, the workshop results, demonstrated a lack of consistent use and definitions of Ra, and did elicit some common use cases. For where this information is used or collected.

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The idea is that this study, design, and workshop, which did include an expert presentation by Dr.

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Popejoy may provide a template for other conferences or organizations to assess their own definitions and Use cases.

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Of Rea. in addition, repeating the study, may increase recognition of the lack of standard definitions and further suggest standard guidelines for collecting this information.

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If you haven't been able to watch the reported presentation yet there is additional data described in more detail.

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There. but i'm looking forward to everyone's conversations and questions.

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Thank you so much, Carly. This is a great overview of some of the important ways in which we use these categories and some of the downstream effects.

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And now we would love to hear from D Mcnight who's a medical affairs director in Vita to see it from the testing company side of things.

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Hi! I hope you can see my slides. Okay, Thank you So I'm here to talk about from the labs perspective.

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How our Ea data is used, and I wanted to first start off and talk about the life cycle of a patient sample or patient case in the lab, and where this might be used.

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We receive the test order from a clinician for diagnostic testing, and the Ra.

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Data is collected on the trf, and I I showed how ours affected on the pre recorded session.

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This is all just noted, put in the systems. The testing was selected by the provider and

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The the order proceeds. So at this point Ra data, provided I a Trf is not used in any decision making by the the lab.

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The sample is then processed and this means Dna is extracted.

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The testing is completed, and there's quality control measures

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I. Our laboratory uses sex at birth as a Qc.

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Metric does not use ra data also at this point so this is not anything that is involved in decision making during sample processing during the after the sample processing the data is generated.

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And it's analyzed where you might see race ethnicity or industry.

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Information come into play would be first in looking at the highest frequency of any variant in a nomad subpopulation.

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So this is Seeing a variant is at a higher frequency than we know that disease would exist in any population. so that is helpful in providing the 9 evidence for a variant and really useful for classifying variance of

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i'm starting significance to benign if if they're too frequent in a population, and you wouldn't expect the disease to be that frequent, or we might see it in the literature in descriptions of a

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founder mutation. But at this point whatever was provided as the patient, what the Ra.

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That was provided for that patient. This is completely agnostic of this data analysis process.

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So, no matter what we're, told or not told for the patient the information is collected from the biomedical research, and it is applied to that variant, and it's completely agnostic of what the patient provided that

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information to us. So so ra! is not used in any decision making for the patient at this point, and then at the very end of report, is generated

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And release to the clinician you might see a mention of Raa.

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If there's a carrier test and there's calculations of residual risk.

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But again, this is a standard information that's provided to anyone ordered the carrier report, and again not not completely agnostic of whatever are you was provided for that. patient.

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So this is just a standard based on literature. that would be reported, and there might be so.

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That might be the only mention. Or, again, if there was literature on a founder mutation that might be mentioned.

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But the patients reported Raa does not determine any of the that decision making, or what's included in the report at that stage.

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So where is it used in the lab because it's not used in the lifecycle of a patient's case.

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Well, there's a few instances where it's used in in billing, and this has come up already in our discussions.

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There's a specific cpt code where a carrier testing can be used for Ashkenazi jewish individuals, and then we know of at least one insurance company that asks to for their patient if they're classified

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as Ashkenazi, Jewish and then that's in the approval of hereditary breast and novarian cancer.

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Testing This is not something we'd agree with we think that anyone should be able to get any testing that that they need for for their their medical care.

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But unfortunately these are sort of some constraints in our systems that that we can talk about, that.

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We are asked to provide that information. In some circumstances. we can then also use that information to ask questions around access and equity.

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So if we look at a geographic region, are we meeting the known diversity in that region?

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If not, is there something we can do about that, or asking questions, or variant and class classification and the equity issues?

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And we know we have problems that individuals from underrepresented populations where they have not been represented in the biomedical literature in our databases have higher rates of the uses.

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So once we track this, we can try to put in solutions to improve these problems, and then we can watch and see if we've made any improvements.

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But we need this information to ask those questions lastly just due to the sheer volume of testing that diagnostic labs.

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Get It's not uncommon, not only for us to have gene discovery, but really to have founder mutation identification, because we could be testing huge numbers of individuals from otherwise underrepresented populations in the

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literature, and so I I know I, in my own experience, have identified founder mutations, and when I talk to other genetic my other friends and geneticists and colleagues, they have examples of identifying founder mutations

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as well. but I just wanted to be very clear, like we recognize that this is like a sociopolitical identifier.

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It is not scientifically sounds so it's not used in any scientific way.

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It is we know there's issues with a self-reported Ra.

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That's why we we take it pretty lightly it's not used in quality control for a patient sample.

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Because of these reasons, self-reported ancestry can be very subjective to depending on where you live.

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And also we know that there's mismatch and and people may not be aware of what their genetic ancestry is also just.

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I think it's important to remember that because all of our testing is ordered by clinicians, although we call it self-reported.

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Information. it's actually physician reported it's not self-reported in most cases.

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So we're currently running this study to even see how concordant

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This physician recorded information. We have matches when we actually do have patient reported information.

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And I also wanted to note that that a significant proportion of individuals leave this section blank, which is, is fine.

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This is not a required section to complete the testing or they choose other, and we'll write in unstructured free text, which we just don't really have the ability to structure that and and use it in in any of

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our analyses at this point. Thank you. Thank you so much to all of our speakers.

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This is really given us a lot of information to think about.

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And and i'm glad to see everybody already including conversation in the chat window.

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What we'd like to do at this time is invite everyone to participate.

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I do have a few questions to get us started while questions are coming in.

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I invite you, as we mentioned at the top, to either raise your hand user, raise hand function, or to type in your question into the chat.

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If you would rather have your question. posed anonymously you can send it to me as a direct message, and i'll rephrase it for the group.

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So anyone who's able to turn on your camera we'd love to see your faces and i'm gonna go ahead and get started with one question for the group.

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The first question being, What are the implications of the observation that Raa descriptors are more frequently solicited on us?

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Requisition forms relative to our international peers. Anybody like to kick us off with an answer to that

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I think, from from our perspective. I mean I I think there's a little bit of following suit.

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So like. This is kind of information that is collected by nih

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Other sort of organizations that that we we kind of follow their lead.

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But then, in yeah, I I asked that question I wonder too, I don't have a good answer.

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But I think another possible reason again is around the billing and and in some cases we can't get paid for testing or a patient wouldn't be able to get testing if if they didn't, provide that information so maybe a

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somewhat of a product of the Us. billing system

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Right. So, having to do with the fact that we have private pairs involved in the mix, which is not always the case.

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Compared to other countries. It came up in Julia's work, and Carly's work as well do you.

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Would you guys like to add anything? Yeah, I was just gonna mention that to. I think.

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You know one of the implications that I have sort of thought about is that you know.

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It definitely shows that it is inconsistent, worldwide, and sort of how we are collecting this information.

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One of the presentations had stated that, you know, international countries are utilizing

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Fill in the fill in the blank or open ended options more frequently than a multiple choice structure.

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So I think it's definitely showing that it is inconsistent worldwide, and also you know not all of the descriptors that we use in on our forms transfer over to our international peers

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Yeah, . i'll go ahead julie please Oh, yeah, Sorry I I was.

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I wanted to add on to that. It was actually interesting, because, another part of this analysis, in another purpose analysis, we were trying to see for those Us labs that had.

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That we're using similar requisition forms in in international settings, and in the Us.

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We were seeing how sometimes, they were trying to be to keep them the same.

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But sometimes it would change them to without them, more to the international context, and in a way, and and maybe they would give more granularity.

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An insert in their populations, and and I guess it's also a question to to answer.

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Do we want to have the same options for population descriptors?

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Or do we want to be more specific depending on who who we're asking, Yeah, Yeah, thank you.

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So much, Juliet, I was I was gonna comment to that it's come up in the chat a bit that some folks are proposing the idea of using kind of a hybrid approach where you have multiple choice to begin and follow up

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questions that are more open-ended. But I have to wonder, then, I, thinking about these comments, where people sometimes check other, and then write in things into the free text response that data just kind of disappears into into the background.

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I saw I At 1 point the Nikita had her hand up.

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Would you still like to ask a question mainly addressed by others?

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But you know I was just kind of thinking. I think somebody asked me to mention this also in the chat just now, but I I kind of wonder how utilized, known as is from the lab perspective. The hearing curation is based particularly on the

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us population. How frequently that data is being used by these international labs, where they may not find it useful to have any proxy for genetic ancestry in terms of their variant interpretation to us They cannot ask about

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it. Yeah, I think that connects a little bit to bob cook Dean's question where he asked how to categories from other jurisdictions map to the Us.

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Categories. How do you manage that? And how do you map your categories to be Sometimes weird population labels in various databases?

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Oh, yeah, that's that's where I saw the link call.

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So yeah, would anybody like to comment on that?

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These questions always make me think of angela samy's work, and then her in her book, Superior.

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She talks about how her South Asian heritage depending on what country she's in is viewed as either black, brown, or white.

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So if you're you know nothing's changed about her she's the same person, but the same background.

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But the cultural context indicates how she's viewed So that definitely tells us there's going to be differences as we move from one country to another, and thinking about how people are going to respond to these questions lakisha would love to hear

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your question. I I also put it in the chat.

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So I think it was carly 1,000 that spoke about that when you asked some about their ethnicity or ancestry, there's a psychological impact. to that.

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So I wonder if you can, you know, speak more to them.

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Yeah, of course. So a lot of our a couple of our participants had shared that there were potentially unintended cycle social implications.

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For when this information may be asked, during a genetic counseling session one of our participants had shared a quote, stating that it might make a patient feel shame or anger for not knowing their

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ancestry potentially for being adopted.

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Not knowing this information a strange from their family. and also potentially for undocumented immigrants that may feel threatened by how that information is used.

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So that was. Those are just a couple of examples of

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What was shared in in my data but definitely something you know from me as a student perspective was something interesting to consider, especially when i'm asking these questions to patients that may not know their ancestry or how to answer the questions it's

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interesting. I ask, because that's the area of my work and I guess it.

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It also depends on the demographics of the studies that you were reviewing.

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You know, setting up the psychological impact a heavy psychological impact for people of African descent, you know, to to answer that question, particularly as a result of champion is late, right?

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And so I was just curious. If you had more thoughts about that.

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So thanks for sharing. Yeah, no problem. Yeah, thank you. And Alice added an asterisk to the answer, Because some folks were wondering kind of what was the group that was participating in this?

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Study, and this was the Wisconsin Genetics Exchange.

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So most of the responses came from genetic counselors in the midwest.

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I see there's a kind of a 2 part question from Mildred that you would like to unmute.

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Yeah, I was just wondering about and you sort of address this in part, but i'm sort of curious for for people who collect data.

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Kind of multiple choice and open ended formats like, What do you end up doing with that?

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Do you sort of throw out the open-ended, or you try to put it into the multiple choice categories, or you just have some other way of analyzing it.

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And then somewhat related to that i'm just wondering in terms of i'm wondering where sort of lab test data on racist missing ancestry.

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Go into an ehr and you know I imagine there's several places where there could be discordance within the ehr and i'm wondering what happens with that I don't know that I can speak to

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where, like a Gen. where genetic lab data would go into the hr I don't have access to that information. but I can speak to, I believe when we get free, tax, we i'm i'm not clear if we try to structure. it I think i've

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seen places, times when we did try to structure it, because we were asking a very specific question around maybe access versus.

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I think most of the time we it it is just not used I think because we don't have really a good way to structure it.

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And so so maybe the answer is that never it never gets used. But i'm i'm not positive about that the only times I I really see mostly us sort of using it.

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Is another place, I guess, would be when we're trying to publish our results in our diagnostic findings from the testing that often the journals will want a a a descriptor of the diversity of the

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population that was tested, and so usually that is in that case I'm.

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I'm pretty sure the other categories just reported as other and anyone who use the the right in is is sort of in just a giant.

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Other category, unfortunately, and then only the structured ones are are usually used.

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In that case

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Thank you so much, De. There was also kind of a related question about coverage, and people were wondering if you could comment on sort of the extent to which these answers influence coverage decisions.

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I I think, for the use of the Cpt code.

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I think, probably pretty significantly. I think we only will use that Cpt code.

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If if we're told the patient is asking as you do is because I do, I don't think it'll get covered otherwise.

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But in terms of the hereditary breast novarian cancer.

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When I checked with our our billing team, I was told it was only you.

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It was only asked for by one payer, and very seldomly so.

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I I think, at least in that instance it didn't seem like it was usually a a major decision, factor, but I I can't speak to when insurance is deciding whether they're going to cover something or not and that

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information's on you know the forms what decision making is happening in in the insurance company that I I don't know.

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I see there were also a few questions. kind of on the variant interpretation side of things, where people were asking about using various ancestral percentages and frequencies to help determine what are the major minor All legal

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frequencies in order to make determinations about variant classification.

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I wondered if Nikita or Kate or anyone else would like to unmute and describe your question or your comments.

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Yeah, I can. I can elaborate if you give me the 2 right it down first to make sure I have it properly, said

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My. My point was that I think, for from these slides I I think she's accurate in saying that it is generally the data is generally agnostic from our classification of there.

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But I think what we do have to remember is that the classification based on minor health frequency, more often than not being more towards the denying side. it is based on the in comparison of the very to what we

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calculate as what's the overall disease that condition, and the way you derive that overall disease oil frequency is from an incidence, number, and then incidents number more often than not comes from the literature which is

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also subject to being based on race and ethnicity, used to define that population.

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And so those metrics for the incidence number for not always something that's generalizable to the whole worldwide population.

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They're again ethnicity specific and whether or not.

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We're using the right threshold there in terms of making more decisions, is a little.

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It's a little bit more complicated and then the other aspect that I wanted to say is especially for those parents that I am suspicious.

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That may be neutral. if I see a variant in particularly one population group in nomad alone versus any other population.

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In my patient when we found this variant is of the same ethnic group.

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I don't think it officially does anything to the variant interpretation, but I do think it's still noteworthy when I'm going through the process and internally.

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I may make a note of that. Perhaps if this is especially in the minority groups.

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Perhaps this is just something that's not well represented in nomad, and later on ultimately figure out that it's probably normal variation in that population.

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So I think there is some utility for getting this sort of information from a labs perspective.

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Whether or not it outweighs all the harm that I'm not.

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Certainly they are not part

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Thank you so much and I think there's another kind of related question, not not specific to very interpretation, but more on the topic of polygenic risk.

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Scores and differential performance in different groups. Could could anyone comment on how Ra.

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Data is used to sort of carry out the analysis for folks who have various, you know, identities, and how the Polygenic risk scores applied or not applied. in these cases.

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I can try to speak to this, but i'm by no means an expert in polygenic risk scores.

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So I I did mention in my recording that we recently acquired a company that was using self-reported ancestry in Pj.

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And a risk determination which we are very quickly changing that to genetic background information.

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Because again, all the the limitations around the self reported ancestry.

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So that that I I did caveat as one place.

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One thing that we inherited that we're working to fix pretty quickly.

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Again. I think that any opinions around prs aside.

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I think that again, most places are using the genetic background as as a way to to sort of population match in that.

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And I I I think that that tends to to be the the more common way to to handle that not self reported. and I think D and yours.

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In your earlier talk you made a an important point which is the self reported, is the shorthand, but sometimes it's actually inferred by you know, Provider's office staff.

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Or it could be any number of other ways of acquiring that data.

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So self-reported is a shortcut that's not always 100%. True.

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Yeah, and It's not just looking at some of that data I could see side by side there's definitely differences in as we analyze it.

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I expect we might even find some trends where we're we're seeing miss reporting at least Miss reporting from, you know, the patient's perspective to the clinicians perspective.

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So hopefully. we'll have information that we can share on that in the near future.

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Thank you, and let's go to anna for her question yeah Well, it was on the protagonic risk scores, and I wanted to know from d like you could think about this in various different ways.

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But i'm not sure it's a good thing if behind this scenes. What you end up doing is partitioning people into these continental ancestry categories, And then, reporting based on that I think we ought to

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try and avoid putting using genetics to put people into these big categories full.

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Stop. I can tell you how polygonic school reporting is going to be done and emerge for which is the biggest implementation.

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Study will be with 25,000 Americans. and behind the scenes.

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We we treat everybody exactly the same. But then, we have this issue that the you do get different performance in different populations, and that's the same whether you look at genetic ancestry categories. whether you Look at race whether you

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look at ethnicity whether you'd like social economic status, whether you look at sex like it varies by all of these different things, because it's not just about the genetics.

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Right there's much. lots of other things going on but it is in, and I like we are going to just states in the fine print.

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What the performance is in different population categories and unfortunately we're gonna have to use these 4 big ones because of details about all the data that we're using to validate the scores

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Thank you so much. I would love to turn it over to Alice.

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Now I see you have some some questions about maybe tools that the group might benefit from using, and kind of getting a straw poll from folks about that

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Can't hear you I don't know if it's only on my end

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Oh, I was double mated, and all for the better because there's a terrible amount of background noise where I am

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But I can briefly say that i'm working on this from a method development standpoint, and would love to hear from folks whether and how a open and the data structure

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Yeah. And Alice had also mentioned in the chat that that Michelle was talking about in the minority genetics, professional network using the open end of question, but not having a lot of uptake on it and Alice had mentioned

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that even the order of the questions matters. if you're using both a multiple choice and a pre response that sometimes putting free response for us can help get more uptake on that.

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Yeah. and how you ask the question. If you say, what is your race?

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Ethnicity and ancestry with an open-ended question.

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People will fill it in. And then, you know, based on the Omb category.

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One, or you know, check all that apply

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Are actively trying to Select, or whether you're just giving it as just like alternative option.

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I think that really matters

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Thank you so much. I also wanna turn it over to Gail.

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You asked a a pretty, I would say, almost full ofical question.

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Would you like to unmute. and share. your question Oh, well, I I think it it's I mean my my my difficulty with this is what i'm sure is.

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Why we have so many people. on this zoom call and that is, that it has never seen seemed to me to have any definitive solution that the utility and that was we you know everyone on this call knows the social construction of

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race and ethnicity, hope, hope, hope against hope, that genetic ancestry might provide a better solution.

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Although Anna Lewis's response was a little disheartening.

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And and so what is the utility? So we see this is a really useful conversation.

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We see this massively inconsistent use across very, very important segments that are trying to, you know.

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Do genetic testing and I I just don't see a solution.

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I think this is always been a problem I mean i've done studies in international studies where you know I've been forced to use the Omb category and categories.

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And it's like absurd it's just I people from Thailand and Africa trying to say, are they African, American or African descent?

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And it's so I I I was hoping someone has a broad solution for us about utility, and I i'm just I i'm not sure what it is.

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I I'm I also wanted to just ask really quickly what respects are to this quite profound chance that the Pediatrics Association in the United States is just the statement they've just put out wanting to prohibit

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race medicine, and I mean I guess I think that's a wonderful effort.

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I think these kinds of discussions tell us the difficulty in using any of these categories as biological or genetic.

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So I wanted, i'd love to hear some solutions to this terribly difficult problem.

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Thank you so much, Gale. This is an incredibly important question. And for those of you who may not have seen the news coming out of the American Academy Pediatrics, This is in reference.

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Yeah, thank you, Mildred. This is in reference to things like race based calculators and things like that, where people use patient, reported race or clinic and determined race, or whatever.

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However, they come to this information to then make clinical decisions using calculators where the values are adjusted.

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Based on these racial categories, and as we've been discussing.

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This is not a biological category, so the use of these calculators deeply, deeply flawed, and has been under the microscope a lot over the past 2 years.

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So any any reactions to this it's really important question posted by Gail panelists or the general audience.

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Nikki does here as a response or a question response.

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Actually . Yeah. So I was just thinking, you know, at least again, because my perspective is mainly live perspective.

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Really the only utility that I really see getting this information is from the very perspective.

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Again. That's my perspective very interpretation perspective is some has somewhat some value, I think. but I don't really need self-reported ethnicity or position reported or you know I don't mean to race and ethnicity.

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I need genetic ancestry so if we can find some. and I think there are people who are potentially working on this lab, or maybe even let me, just to some extent or no man specifically I think we can infer somebody's

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genetic ancestry to some extent from these panels, because we were doing all panels and getting a lot of sniff data on what are just normal variants in there.

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We can infer their genetic ancestry to some extent, probably with some complicated mathematical probability in there that I wouldn't understand. And thus we can get what we need from the very interpretation perspective without actually having to

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ask for that in any official sense, and have it officially documented anywhere.

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And let it just be something internally that the lab folks use I I don't know if that would fix the issues that Gail is alluding to.

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But I think it might be a step in the right to take it out of any official documentation, but still get the better looking at genetic ancestry

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Is that the solution? then? ? I don't know if that's the Solution? And you seem to be speaking kind of contrary to that.

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Yeah, I just think, like the main way the genetic ancestry is thought about is this continental ancestry categories, and those are quasi ratio. And I just put it in the chat But whenever

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we talk about whenever we talk specifically about genetic chemistry, and especially when we need the consonants and sisy categories, we're emphasizing between group biological differences which history teaches this is dangerous thing and I do

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think that genetic answer. She can be part of the solution.

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But we really rapidly need to change course, and to mean by genetic ancestry something different than these continental ancestry categories.

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And I think that's the challenge and the things that we need to do I think there's a way to avoid.

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So you're you're right in saying that these we're mostly using Continental populations.

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When we're using the genetic ancestry in this analysis.

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But there's a way to just avoid using labels at all, like, for example, using unsupervised learning.

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So you know, like supervised learning you're using labels in order to to predict things where it's like unsupervised, you're assuming there's just there's like free for example, 5

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6 7 categories, But you're not specifically labeling them.

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And it's the algorithms that actually group these individuals into these like abstract groups like unnamed groups.

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And then at the end of the day there is a way that you can set those labels after like.

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Look, look at your data and be like, Oh, right like these people are grouped together.

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They're actually from Europe. This people are from this other place But I I ideally, you want to use these kind of methods that are just the maybe are just grouping without a label, and and maybe that that's the way, forward.

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Like, maybe telling these algorithms, give me like 10 categories, or even more, and not actually naming them.

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And just treating these as people there that are genetically similar, but not necessarily part of a group.

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There. you can just use nitp patterns or you can just see patterns in in their day like later on. We won't even be able to have categories at all, because people will just be a mix of of of people you know so

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so that's maybe the way forward

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Thank you so much for your comments, and I just wanted to point folks towards one of Anna lewis's and teams.

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Recent commentary is really great description of The dangerous ways in which trying to use genetic ancestry and continental definitions actually just recount kind of recreates race in many ways as as anna was saying

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So I see we're kind of winding up on the very end of our of our time together.

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So I just wanted to pose an overarching question to the panel.

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That will help us actually transition to the third session in 2 weeks.

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So for the panel, and also just generally for the audience.

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Based on these conversations. Do you feel there's an emerging answer to the question of whether and how to ask about Raa information in context of clinical genetics

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I I don't know if there is I hope there is I I said in my talk, I think we're looking for some guidance.

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I think we'll well do you What what the community asks us to do or collected.

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How the community asks us to collect it we're pretty flexible in that, because again, we see somewhat the limited utility.

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There is some utility, though so so definitely I hope that the community is able to to find a way to to incorporate that.

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I also do not know if there is, you know, a specific answer to this or solution.

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But in the context of genetic counseling you know i've found, at least for some of the patients that I've had that sometimes they may not understand the question that i'm asking them, and so they may be providing their

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race when i'm really looking for their ancestry so just phrasing.

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You know what countries of origin might your family be from

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Or if a patient does provide their race potentially asking a follow-up question, to ask for more specific countries of origin, if they know that information to give them.

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You know that space to consider thinking about that information and and what their country's origin may be

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I had a little come in there, too. So yeah, I guess I agree always with you.

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There is an like a clear answer on this. however, I do think with you know, in the future we will be able to develop methods.

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I hope that that can help us just not use like set labels that are that make people feel like they're put into boxes.

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We will find a solution that makes everyone feel like they belong to a community, but not necessarily a community that has a predefined way of doing things, or or has seen in some predefined way.

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Thank you so much. These are all such important considerations for us to think about, not only in terms of the downstream analysis, but also, you know, the initial patient castling and providing anticipatory guidance about potential

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outcomes for these test results based on what we know about their backgrounds.

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So thank you so much to the panelists, and also everybody else for your engagement. and you know contributions to conversation really excited to keep this going. In 2 weeks.

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The third session is going to be focusing on revisions to demographic representations on clinical lab record.

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Acquisition forms as we've seen there's a lot of flaws, and how things are done now and you know It's a little bit the wild West.

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There's kind of every possible way of doing things so trying to see if we can put forth a few sort of proposals and come to over at some sort of consensus, if possible, in one short hour together, but that

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session will be happening in 2 weeks, and the information was just posted, I think, in the chat about how to register, and there will also be a follow up survey for your feedback about these sessions So thank you all So much for your

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engagement, and it's great to see everyone thank you so much Thanks, everyone.