Excerpt from Question 3 Working group Conference Call 1 9-12-2016

DR. DANIELS: Yes so and we're taking notes too so we'll be able to try to integrate some of this into that discussion as well. Any other comments about the portfolio before we move on to duplication? So the fourth item on my agenda was to ask the working group if you noticed any areas of this question or specific projects that you feel any concerns about related to duplication of effort.

Or if you have any suggestions of how duplication of efforts can be avoided in this area of looking at risk factors, so this is question that we're asking because the new law, the autism chairs act requires that the IACC and its strategic plan provides some recommendations about avoiding duplication of effort.

And so we wanted to follow through on that and let everyone one of these working groups have a chance to take a few comments about this. So does anyone have any thoughts about duplication? Any concerns?

DR. EICHLER: I mean, I can comment because I've seen it. I think the most important thing is to have the manifest of the datasets that are actually being sequenced by whom available to all and I noticed, so we know that in the actual sequencing of autism genomes, there's been redundancies between missing and other efforts.

And that's in part because the data manifest in terms of what samples were being sequenced (unintelligible) and other questions were not made publically available. So I think that what we really need to have is an open dialogue between NIMH the foundations that are sequencing for examples, that's one area, and just making sure that those manifests are in place long before sequencing starts.

And all it requires is kind of a higher level coordination and people can find additional samples to sequence or you know, or not to sequence. If they…

Speaker: That sounds like a great…objective

(Crosstalk.)

DR. EICHLER: Well it's a pain, I can tell you it's a pain because someone has to sit there and actually do this kind of coordination but it in the end saves millions of dollars to do it, right? And so…

DR. HALLADAY: Well you know, yes say it's the cost of a person or a part, a part of a person's salary to do all that, I mean, to make sure that happens, that seems to need a minimal amount of money to make sure it happens.

DR. EICHLER: Agreed. And just, and the only thing you need which is the biggest problem was basically having all the foundations agree that they would share what their sequencing and that has not always been transparent.

And that's, I don't know how you leverage, I mean, just having one person unless they don't have a stick it doesn't make any difference, right? They won't release, they won't release. But I think in the end, everybody wins if people are open about what they're sequencing for example.

But that just, that's been a little bit slow in this area.

DR. AMARAL: Okay, Evan, this is David. I just want to understand what you're saying. So initially was thinking that you were saying that there was redundancy and different efforts sequencing the same subjects and wasn't clear about that.

But then at the end of your comment, it sounded like there was something else but I'm not sure if I understand exactly where you, what you're indicating is the redundancy. Could you try one more time so we understand it, I understand it?

DR. EICHLER: The redundancy is really just what you first mentioned, same samples being sequenced twice by a different competing efforts, right? That's the redundancy.

DR. AMARAL: Okay.

DR. EICHLER: The problem is release of what we call the sample manifest, who's sequencing what? And that you know, centers have been I don't for unintelligible) trend or they necessarily had their manifest all in place. I don't know what the issue has been but that has required a lot of effort to convince people to release their manifest to each other almost if that was a sequence in itself.

So I know NIMH can obviously make all its investigators that are getting funded make it really clear what you know, what samples are being sequenced but if the same samples are being done by missing or being done by you know, by the Simons foundation or something to that effect, that's not always clear.

Nor are those places obligated to share those, that information. So that's a separate issue so that's what I'm saying it's not just having you know, one FTE focused on you know, coordinating or even half an FTE to do that. But it would be nice if there was just one clearing house where we knew all the samples that are being sequenced by whom.

And I predict that would save money and lots of it but does that make sense (David)?

DR. AMARAL: Yes, no, it does make sense. So what you're asking for is more of a formalization because if understand correctly, I know that Spark is declining subjects that are being sequenced by missing you know, at least that's what they tell people who are involved in the studies.

But you're saying that may not necessarily take place and even if it does that it would be better to formalize that and publicize the, as you say the manifest of what they're doing? So I think that's a good point and I just wasn't aware of how problematic that is but if you think it's a big issue, I think that it's something we should address more formally in the next go around of this.

DR. EICHLER: And I think this is also totally intertwined with data access because if you have to pay Google for example to access missing, a huge amount of money, right, that will exclude most investigators from actually analyzing that.

So some colleagues have said you know what? We're sequencing the same sample but to be honest, we're going to make it publically accessible so that data can be accessed and downloaded and as opposed to having to actually maneuver through you know, the fee structure of actually you know, analyzing or manipulating data on the cloud.

So these, these are complicated topics to be honest. It sounds simple, share manifest, but it's also important to know you know, how accessible is that data going to be and to whom and how much is it going to cost.

And this is where I think the institute or NIMH in particular should be playing a big role because like I said before, it benefits a lot of people if all this data can be put on one platform and made freely accessible at least to qualified researchers.

DR. AMARAL: So I do think it would, it's under the purview of the committee to recommend that some funding be allocated for coordinating meeting genetic data that is coming in through various sources that are relevant to autism research.

So if it isn't being done know, you know, I think it's something that when we get to the third call where we'll be talking about new goals or maybe this is actually better for Alison's, you know, in question seven.

But somebody should say that some funding should be allocated to bring all the critical parties together and solve this problem so that you know, so that you and other investigators who are dealing with these kinds of data don't feel that access isn't open and rapid.

And you know, it shouldn't be as costly I mean, I think that this is really an important point and I don't think I was aware of the depth of this issue so we should certainly keep it on the, on the front burner.

DR. EICHLER: Yes. The good news is that the data coordinating centers from the CCDG at NHGRI are beginning to do this for you know, a little bit for autism but for many of the other diseases that are going forward for genome sequencing.

So it's starting to happen but it just needs to be I guess coordinated I guess is the point, coordinating the coordinating committees I guess.

DR. AMARAL: Yes. Good point, (Evan).

MS. SINGER: This is Alison and I also wasn't really aware of this. So I do think this is exactly the kind of thing that we include in chapter seven. So I think what would be great is Evan is if you could just maybe shoot me an e-mail with a couple of sentences about this and I can bring it up on our chapter seven call.

DR. EICHLER: Okay I will do that. I will probably send it to you in a couple days. I'm sorry I have to run guys but this has been an interesting discussion. I'll talk to you guys later.