Adrian: [00:04] Hey, this is Adrian Hernandez and welcome to the NIH Collaboratory Grand Rounds podcast. We're here to give you some extra time with our speaker and ask them of the tough and interesting questions you want to hear most. If you haven't already, we hope you'll watch the full Grand Rounds webinar recording to learn more. All of our Grand Rounds content can be found at rethinkingclinicaltrials.org. Thanks for joining.

Lesley: [00:28] Today we're here with Andy Faucett who'll be reflecting on considerations for the return of genomic results. Andy, it's great to have you with us today.

Andy: [00:38] It's great to be here.

Lesley: [00:40] You know, I want to start with a question around some of the ethical and regulatory concerns with return of genomic results. What has been your experience in dealing with local institutional review boards and how they think about this important area?

Andy: [00:59] I think it's a growth period for IRBs. I think that they also, it's unfamiliar territory for them. So one of the things we've done at Geisinger is we have genetic counselors and/or geneticists on our IRBs. We're constantly doing education. We've also progressed over time. Our initial consent form didn't allow for return of results.

Andy: [01:20] We then did focus groups and took that evidence back and had a broad discussion with our IRB. So I think it does take discussion and it is a time process. We're all learning. I think years ago, we were afraid of genetic information. Now we're finding that people deal quite well with it, so maybe our fears were misplaced.

Lesley: [01:40] Yeah. When you first started this, what were the conversations with the IRB like then, and how have they changed?

Andy: [01:49] Well, it was partially IRB, but it was even where genetics was. I mean, when MyCode started in 2007, the assumption was that you wouldn't return results, that you definitely wouldn't put them in the medical record. I think what we've learned over time is that you want them in the medical record because that's how they're going to be beneficial, if you're returning beneficial things. You need to be careful about what you return, there's some things you probably don't want to return.

Andy: [02:13] But again, it's that conversation that I think has really moved us forward, not only in genetics but also in the IRBs because I remember in the early days when you did BRC1 or BRC2 testing, you had to come in twice for genetic counseling before they drew your blood and now we know that sometimes you can have that conversation over the phone and do testing. So I think we're all getting more comfortable that people deal well with the information, and that the risks are pretty low.

Lesley: [02:41] Yeah. You know, you mentioned there the benefits associated with having some of this information in the medical record. I know certainly we hear a lot about concerns with how these results might lead to discrimination, which is not a benefit of having them there. Can you talk a little bit about your experience with those concerns?

Andy: [03:04] Yeah. It comes up in most of the focus groups and some people ask about it on a regular basis. What I kind of hear is if you're focused on conditions that are medically actionable, most people say, well, if I take the potential benefit to my health and my life by knowing the important information versus the risk of insurance discrimination, I'd rather have the health benefit, and then we do have lots of things in place.

Andy: [03:29] Now we're not returning results that have no actionability. We're not returning things like Huntington's or Alzheimer's because at the moment there's nothing to do with those. So again, I think that helps people with that.

Andy: [03:42] One interesting side note was that in the focus groups, just by random selection, we often had one or two healthcare providers in each group and they tended to be the ones that were the most concerned. And it makes me wonder if those of us in genetics and in healthcare have been over-sensitized. That the general population clearly said, if this is something that might benefit me, I'd want it in my chart.

Andy: [04:04] And then in relation to that, you know, we've been returning results for about two years and we have about a half a dozen cases where we've either found cancer or heart disease early that would not have been detected without the individual having participated in MyCode. So we're starting to see some pretty, pretty important stories that we can share.

Lesley: [04:24] Yeah. Now I'm curious, how do participants deal with where we are on this learning curve? Because you pointed out certainly in your presentation that this is a learning curve and what we know now is so much different from what we knew in 2007 or even a few years ago.

Andy: [04:44] One of the hallmarks of the program at Geisinger is that we've always tried to be as transparent as possible. And in the focus groups we talked about the fact that our knowledge today was limited and that's why, you know, humans have 20,000 genes, but we're only going to look at a hundred or so. And people said, not surprised. We understand this is a new area. There's a lot you're not going to understand.

Andy: [05:04] So I think, at least in our experience, the importance is being very clear and transparent about what you know and what you don't know and that we have a lot to learn and that part of the reason we want to do this research project is so we can all learn together.

Lesley: [05:18] Do you, do you hear concerns about delaying, for example, how often or how quickly you share those actionable results with patients?

Andy: [05:30] Yeah, we have. One of the issues we run into is that some people are like, well, I'll participate in the research and I'll put off doing clinical testing and we strongly send out a message that's not the right way. First off, because our test, our research test, isn't testing for everything that's needed in clinical.

Andy: [05:48] But we do have to remind people that this is first and foremost a research project, and we're on a research time schedule. We like to do sequencing in large batches for quality control and being able to look at batch effect and those kinds of things.

Andy: [06:00] So it is an ongoing discussion. But again, I think if you're, you know, if somebody, usually I find the people who are worried about how quickly the results come back, have some reason they probably need clinical testing and sometimes that discussion will bring that out and then we're able to get them into the clinic and make sure that they get the test they really need, versus someone who's truly just participating and doesn't have something in their family they're concerned about.

Lesley: [06:26] Andy, any other sort of highlights that you'd like to share with our listeners about your work and what's coming up?

Andy: [06:36] I think, I think over the next two or three years, we'll be able to show that there may be some conditions that we all should have genetic testing for. We are talking at Geisinger about making exome sequencing something we offer all of our patients. I do think that day is coming. I think we're certainly ahead of the curve.

Andy: [06:57] And then lastly, since this is kind of a research focused talk, I think that the old model of research subjects and that I'm going to take things from you and not give you anything back is just not okay. I think it's research partnerships and we're actively trying to work with our research participants involving and making sure we're asking and trying to answer the questions they have. So I think the needle's moved and I think saying you're not going to return results is really not okay in today's world.

Lesley: [07:28] I agree completely, Andy. Thank you for that. Well, and thank you for today's discussion. This has been terrific. I want to highlight for our listeners that our next podcast will be Ellen Tambor and Dr. Sean Tunis on straight from the source, clinicians' views on participating in comparative effectiveness research and patient-centered outcomes research.

Adrian: [07:57] Thanks for joining today's NIH Collaboratory Grand Rounds podcast. Let us know what you think by rating this interview on our website and we hope to see you again on our next Grand Rounds, Fridays at 1:00 p.m. Eastern time.