

- Speaker 1: From the National Society of Genetic Counselors, this is the NSGC podcast series, exploring stories of leading voices and best practices in genetic counseling. Now to your hosts, Khalida Liaquat and Kate Wilson.
- Kate Wilson: Welcome to the NSGC podcast series. I'm your host, Kate Wilson. Today, we are partnering with the Journal of Genetic Counseling to further explore two articles featured in the journal. First, my co-host Khalida will talk to Nicole Bertsch, co-author of "Perspectives on facilitating whole exome sequencing for international patients at Mayo Clinic," and later, I'll speak with Jenny Morgan and Patricia Birch, co-authors of "Indigenous Peoples and genomics: Starting a conversation."
- Kate Wilson: Each individual joining us today will help us to understand the importance of working with diverse patient populations. Take it away, Khalida.
- Khalida Liaquat: Thank you so much, Nicole, for joining me today. Nicole, you wrote an article for the special issue for the Journal of Genetic Counseling titled "Perspectives on facilitating whole exome sequencing for international patients at the Mayo Clinic," and I was really interested in knowing a little bit more about the work, the study, and how this topic came about for you guys.
- Nicole Bertsch: Certainly. Thanks for having me, Khalida. When the Journal of Genetic Counseling sent out an email asking for articles about whole-exome sequencing, we as a group of clinical genetic counselors thought this could be a good opportunity to fulfill a professional goal we had of writing a peer-reviewed paper since facilitating whole-exome sequencing or WES, as I'll call it from now on, occupied a large percentage of our clinical time as counselors.
- Nicole Bertsch: We started to reflect on our general experiences with WES, some of the challenges and opportunities we had, and our experiences specifically with our international patients kept resurfacing as some of the most challenging and thought-provoking for us in terms of reflecting on our skill set and our abilities as counselors and just kind of some of the more memorable sessions had to do with that population, so this is where we figured we'd learn the most and we would try to share through the Journal of Genetic Counseling.
- Nicole Bertsch: The aim of this article is a commentary about our experiences, and we really aimed to start a dialogue with other genetic counselors and providers and other clinics about their experiences facilitating WES for international patient populations, and how we defined that was a patient coming from another country to Mayo Clinic for a medical evaluation, and for us, that often looked like the patient being initially referred to another department, such as cardiology or neurology or pediatrics, and then eventually being referred to genetics for a full workup, which then if the geneticist deemed inappropriate would include whole-exome sequencing.

Nicole Bertsch: The article goes on to describe the general process that's in place at Mayo Clinic for facilitating WES and how the genetic counselors are involved in both pre-test counseling and then on the back end once results are available, presenting them at departmental meetings, and then finally post-test counseling, so we're essentially point people throughout the entire process, and then the next sections describe the different characteristics of our international patient population, and our patients at Mayo Clinic mainly came from Middle Eastern countries.

Nicole Bertsch: And then we divided the challenges that we faced with these patients into two main categories, the first one being logistical challenges, which was further broken down into facilitating in-person results disclosures since these patients' visits are very limited, and their ability to go back and forth is kind of a one-time thing, and then the unavailability of samples for trio whole exome sequencing, that if we only had one parent and the proband present, then we weren't going to get that other parent, and then challenges with just general follow up and medical management once they return to their home country.

Nicole Bertsch: And then the next category of challenges were more specific to counseling challenges such as managing these patients' expectations for whole exome sequencing and countering potential stigma of carrier status and also cultural norms regarding the carrier testing process and also reproductive options that we hadn't necessarily encountered before. We summarized those counseling challenges as being very difficult in the context of whole exome sequencing because of just the sheer amount of information that's conveyed during the pre-test counseling session, and then the reports on the back end are pages and pages long of a lot of information, so there's just a lot to sort through. Then there's time constraints, language barriers, and then a big emphasis on education too, so we found all of those to be very challenging. And then finally, we end by discussing different approaches to multicultural counseling, and we suggest different resources.

Khalida Liaquat: Sounds like a really well-rounded article. I have so many questions. The topic of inclusion and diversity has been certainly on the forefront of a lot of our minds in the recent times, and this is a very specific population. It's international, it's Middle Eastern, and it sounds like all of you collectively learned quite a bit from your experiences here. Is there anything that you found surprising or particularly challenging when you reflect back on the data you're presenting here?

Nicole Bertsch: As we reflected, it was interesting to us to see actually how many patients we were able to give in-person results to, so at first, we were like, "Well, is this really a challenge? It appears that actually most people got their WES results in person," but then as we reflected, we realize, well, a lot of those in-person results disclosures came with some added stress of, "Oh, the family is leaving for Saudi Arabia tomorrow, and WES results just came back. Where are they? How do we get a hold of them? I already have a patient scheduled this afternoon. I

need to move them off my calendar so I can see this family. They're taking priority," so some of that, we didn't really necessarily elaborate on a whole lot in the paper.

Nicole Bertsch: But then it was rewarding to see in the quantitative data that actually, a lot of those methods that we put in place, like the service delivery model that we described in the paper called Genetic Counselor of the Day, did appear to be working, and obviously we can't attribute everything solely to that, but it was rewarding to see that coming to fruition with some of our numbers that we'd put in the paper. But some of the challenges that we had are still reflecting on. In our very last section, we talk about next steps that you can take, and a lot of that has to take place outside of the actual counseling session, but after you see a patient, you have documentation to do and other follow up, and so it'd be ideal if you had an hour every day that you could devote to reading papers about clinics in other parts of the world that do genetic testing and work with these patients. We just don't necessarily have that time, so there's also a lot of talk right now about how do we streamline workflows? How do we become more efficient to allow for that kind of being a continual learner?

Nicole Bertsch: It was very humbling to see, how can we prioritize this as a team? Right now, it just seems pretty difficult to do, but it's something that we really wanted to make a priority. And then the final challenge that we note in the paper and continue to reflect on is the lack of follow up and management that results for these patients as they return to their home country. We really have no idea if these results are getting into the hands of providers there back home. We don't know if these families are getting connected with genetics back home, so as we as a field try to measure things like cost effectiveness and genetic counseling outcomes, that's really difficult in this patient population, but it's a large percentage of the population that comes to Mayo Clinic. We don't have really an answer for that, but that was one big challenge that we continue to reflect on, and hopefully, moving forward with this paper, we can start dialoguing with other clinics that are also large medical centers and maybe see some of these patients as well and get some of their thoughts and approaches.

Khalida Liaquat: It sounds like a really good opportunity for the GC community who maybe struggles with the same issue of international patients and follow up to get together and find best practices.

Nicole Bertsch: I mean, we can't be the only ones, so we just wanted our thoughts out there.

Khalida Liaquat: And then the other thing that's interesting to me, especially with respect to communicating with other GCs who deal with international patients, is you mentioned this is a particular subset of population from the Middle East. I wonder if some of the challenges you have with this population are similar to others maybe who see folks from other parts of the world, and so with that in mind, this could go into a lot of different directions for further publications or

other educational opportunities. Where would you like to see this particular work that you publish take a step further?

Nicole Bertsch: I think it would be really fascinating to do more of a qualitative analysis of these families' experiences. When they travel to the other side of the world to seek medical care, to us when we meet with them and discuss WES, their expectations are, "I'm here. I've traveled all this way. I've put in all this effort. We're going to find an answer," and so part of our role is to set expectations up. WES is extensive, but it's not exhaustive. There's no guarantee, and that can be really, really difficult on the back end for families to comprehend.

Nicole Bertsch: And even if they're local to the United States, it's still hard, but I think especially because of the extra effort and just that mindset of, "Oh, we're going to come to one of the top medical centers in the world," that's how they perceive Mayo Clinic. "We're going to get what we came for," so I would really like to be able to sit down and say, "Where are your expectations coming from? What has informed them? Is it the media in your home country? Is it just what you've read or what your doctors have told you about what genetics can do? What is influencing that?"

Nicole Bertsch: It's also talking generally to them about, "How has your stay been here? How has it been being in a foreign country for three to four months while your child or your family member is really sick, and you're hoping for an answer?" Exploring that even further, and then ideally, we could try to establish connections with these families on the back end once they return to their home country, say, "What did you do for follow up? Did you get to connect with the genetics clinic there? How can we help facilitate that?" To again try to measure more of those outcomes of how effective we are, and even just thinking of WES globally, increasing that access and making sure that making a diagnosis through WES is effective.

Khalida Liaquat: You describe logistical challenges and counseling challenges obviously with a multicultural aspect, and taking a step back from that, do you feel that you and your peers were well prepared to tackle some of these challenges, either based on your experience as clinical counselors or through training? Are there opportunities there that were noted and maybe didn't make it to the publication?

Nicole Bertsch: That's a good question. I would say we would all say we were fairly well prepared to use an interpreter. I think that was pretty standard in all of our graduate school training, and we had some experience using interpreters for other types of counseling, so I would say interpreters in-person or over the phone or using an iPad was all pretty standard, but I think beyond that, we felt like we lacked just resources and understanding where to go to learn more about Middle Eastern culture, or how do they view different health issues?

Nicole Bertsch: The more we dug into that though, we realized that at Mayo Clinic, the HR team offers multicultural seminars, or there's even a website you can go to that you can click through different ethnic groups and learn more about them and specifically views pertaining to medicine that might be pertinent, so we're like, "Oh, wow. That's really good to know about," but we kind of had to dig for it. But once we learned about that, we felt more prepared.

Nicole Bertsch: The other resource I'll mention that I've been vaguely aware of as a student but haven't really looked into it is Transnational Alliance for Genetic Counseling, TAGC. I believe right now, it's actually free for genetic counseling students to join, and this appears to also be potentially a great way to be building connections with genetic counselors across the globe and, like you've been saying Khalida, starting this dialogue and helping it continue further, so that's another resource I would point out and we were very happy to learn about.

Khalida Liaquat: I wasn't aware of that group. I think for any counselor who is in any way related to populations that they're not familiar with, it sounds like a good resource to just start a dialogue or get more information prior to seeing patients. Nicole, the publication's coming out in the special issue. Do you see this being something that you're going to continue to publish in? What does this look like for you?

Nicole Bertsch: I would definitely like to continue to be very intentional in learning about patients' cultures, whether that's during a session itself, taking time to say, "Tell me about some of your family traditions," even as you take a family history. If they bring something up like that, really exploring it, and using TAGC to continue to make those connections, to just not let my time be wasted as well. When I find myself with some down time, perusing Journal of Genetic Counseling or even PubMed for newer articles, so just continuing to read.

Nicole Bertsch: And I think too, realizing that with patients who are from a different culture than you, it might be more difficult to relate in a way that you don't necessarily gravitate to, like you might not be able to say, "Are you glad that Virginia on the NCAA tournament?" You might have to start on common ground of, "It's natural to fear for your child's health," and that can be a little bit uncomfortable, but finding that common ground in an area that isn't maybe a natural way to start off contracting, but you have to remember that everyone's just a human being at the end of the day and finding that commonality and building from there I hope to keep a priority.

Khalida Liaquat: And what I often tell my students is the parents of a sick child are the parents of a sick child no matter where they're from, and so there's always going to be that commonality that is there as a clinical counselor.

Nicole Bertsch: Right. It's just recognizing that and being okay with starting with that commonality and saying, "I understand that you're here because your child is really sick. How has that been for you?" I don't know if that's always our most comfortable to approach a family, but we're trained to do that, and we

emphasize in the commentary that we as counselors need to not let our skills get lost in the shuffle, especially with WES, just the information overload that comes with this type of testing, but still be engaged with our counseling skills and not lose that part of our title.

Khalida Liaquat: I love that phrase you used, still being engaged with your counseling skill no matter how complex the genetic testing has become or will become in the future because I can only imagine the Mayo Clinic continuing to offer complex testing to wider populations because as you said, it's a world-class facility. Genome is next, and who knows? I think this publication is important stepping stone. I think it's a really good dialogue-starter, and I'm really looking forward to seeing what comes of it, so thank you for coming up with this idea, you and the group.

Nicole Bertsch: You're welcome. Thank you for having me, Khalida.

Khalida Liaquat: Check out the Journal of Genetic Counseling to read more about Nicole and her team's research. Next up, my co-host Kate Wilson will sent down with Jenny Morgan and Patricia Birch.

Kate Wilson: Thank you for joining us today.

Patricia Birch: Thanks very much.

Jenny Morgan: Hi there.

Kate Wilson: First up, I wanted to see if you could tell our listeners more about the study that you did and the publication in the Journal of Genetic Counseling.

Patricia Birch: The goal of our study was to raise awareness and to give voice to indigenous Canadians because they have lack of representation in genomic databases like NOMAD, and that has consequences to their healthcare from an equity perspective, so that's the really quick synopsis. And it was actually one indigenous family in particular that was the impetus of the study, and it's probably a pretty good illustration, so they had genomic sequencing in a hospital, and a genomic variant was found in their child.

Patricia Birch: And it was an incidental finding that hadn't ever been seen before in the various databases that we can use, but its location made us worry that it could be quite serious, but on the other hand, it was also possible that the variant is just a normal, common variant in people of that particular indigenous ancestry, and that sort of highlights the fact that the problem is that the reference databases like NOMAD lack Canadian indigenous genomes, so we couldn't be sure of the interpretation, and that lack of diversity means that the genomic healthcare is compromised for about five or six percent of the Canadian population who are First Nations, Métis, or Inuit. That's the problem in a nutshell. I think Jenny

would like to relate some of that to the Truth and Reconciliation Commission in Canada, especially for non-Canadians.

Jenny Morgan: In 2015, the Truth and Reconciliation Commission published a final report with calls to action, and the work they were doing was looking specifically at the legacy of the Indian residential schools in Canada and identifying collectively from indigenous peoples what some of the issues and concerns were, but what were some of the solutions to that? And so when we look at the calls to action, we can see what are some of the pieces that we can implement into our work that we do? And in addition to that, the TRC Commission published the principles of reconciliation and what does that look like, and the very first principle is the use of the United Nations declaration of the rights of indigenous peoples as the framework for reconciliation.

Jenny Morgan: That's always at the forefront in the work that we do when we're prioritizing reconciliation is ensuring that the rights of indigenous peoples are upheld in the work that we do, and then that gets back to some of the concerns that this study is raising around the inequities that indigenous peoples are facing.

Kate Wilson: And I think that you both bring up a very important part and something that I noticed in particular about the article that you had in the journal. You talked about giving a voice to indigenous Canadians, and then Jenny, you're talking about using the Truth and Reconciliation Commission as well as United Nations and making sure that the rights are upheld in the work that you're doing, so I wonder if you can talk a little bit more about how you ensured that those rights were upheld and how you included indigenous Canadians into the study design?

Jenny Morgan: Sure. Right at the very start, when we were composing the team, we knew it was imperative that indigenous peoples be a part of the process from the start, so we ensured that we had indigenous representation within the research team, and we participated in the design of the research. And when we're looking at some of the methods that we used, we knew that we wanted to incorporate some mainstream approaches such as focus groups, but looking more specifically, what does that look like from an indigenous approach?

Jenny Morgan: We used what we call sharing circles, and we had an elder involved who was also part of the research team who helped facilitate the sharing circles. Part of customs is sharing of food, so just making a very safe, comfortable space for indigenous peoples to gather how we would traditionally gather together, and just ensuring respectful exchange of ideas and creating, again, that safe space for indigenous peoples to be open and honest with what their concerns were, so just ensuring indigenous peoples were a part of the process right from the start to the finish, and in the design as well as later when we talk about the results that came out of the study.

Patricia Birch: This is Patricia here. The analysis was carried out by a team. Those of us of European ancestry and those of us of indigenous ancestry, we did it together,

and then the important part I think also of the method was the fact that we took the themes back to the participants, and we discussed them with them, and we showed them what we had done, and we asked for feedback on it. And we did receive some feedback, so that is also part of the trust-building, the opportunity for us to work together with research, which is somewhat unique for most types of research, that's a research model that we call community-based participatory research design, and we actually think it was a pretty good framework for this purpose, and it's certainly congruent with the genetic counselors' collaborative practice, so that design would be something that we think would be worth exploring for anyone interested in this type of research.

Kate Wilson: I found that was an interesting design, and like I said, it was something I had not heard too much about. I think one of the things looking through the work that was done was the thoughtfulness that you had and the awareness that you had, and not just the design but like you said, at the end, going back to the participants and discuss the things. Was there anything in the course of doing the study that you found particularly surprising or challenging?

Patricia Birch: I think in terms of challenging, one of the biggest challenges was that the funding agency put a one year time constraint on the project, and that's something that was not really sufficient for this type of a model of research, particularly with indigenous people, because it didn't allow us enough time to engage with indigenous leaders who are external to the team, so we couldn't capture their viewpoints. Do you want to say something on that, Jenny, too?

Jenny Morgan: Sure, and I would say more broadly, when it comes to research with indigenous peoples, researchers have to take into account the history and role of research with communities, and if you look into a lot of what indigenous scholars are writing and have written, they really go into detail of colonialism and the tool of research and what it's been doing with communities, and we wanted to ensure that we're not replicating that in the work that we do, and so there was still a lot of distrust because of that history with communities.

Jenny Morgan: And so a lot of communities have now put into place research protocols based on past experiences to ensure that their communities are represented in a good way and the research that's happening together with them rather than on them, so we just had to be aware of the uniqueness of different communities, where they're at with research protocols, and our understanding that we need to abide by some expectations that they have in order for them to also participate with us.

Patricia Birch: In terms of things which could be thought to be surprising, given the horrendous history of genetics research in North America and particularly at our university, where there is a shocking case that happened in the 70s and dragged right the way through to nearly 2000s, but all that being said, the 30 participants in this study in four focus groups were from a variety of places, and they were

incredibly engaged, very perceptive and thoughtful, and came up with some very profound ideas.

Kate Wilson: And I think that that was one of the things too, seeing the discussion that occurred and that you had pulled quotes from some of the participants in the write-up, which really helps everyone understand the lack of trust as well as some of the history with the indigenous communities in research. Was there anything additional that you worked to do to try to build that trust or if there was anything else you found that was helpful in working with the indigenous communities or anything potentially you might have tried differently had you had a longer time to work on the study?

Jenny Morgan: Within the focus groups themselves, I think a major piece was having the presence of the elders, so the elder did a land acknowledgement and welcome with the participants, and that was very important to creating that safe space for participants to feel so open because we are asking them some information, and I think as researchers, we had to also be aware of some topics that might be either triggering or challenging for those participating to discuss, especially when we're looking at ancestry and some of the challenges or even misidentifying that as being ancestry testing, so we had to consider a lot of things that might come up.

Jenny Morgan: And we had conversations as a team, and even amongst the researchers and team, the elder was very important to help prep us before moving forward with the participants.

Patricia Birch: We also had described during the focus groups who was indigenous, so generally speaking, I think that combination of things, the sharing circle approach, the food, the elder, really helped to create a safe space.

Jenny Morgan: I would also add that within the focus groups, we really did our best efforts to go out to the community, so we identified some locations where the community members were coming from, and so it was us coming to them, and again, that was a part of being in a familiar place for them where they felt comfortable to reduce some of the barriers to participating as well.

Patricia Birch: One of the other things that we didn't talk about was that we had to somehow create a common body of knowledge about genomic testing and the issue of non-representation of indigenous Canadians in things like NOMAD, and we had to do that quickly and efficiently, so the five minute introductory part of the video that explained that, we took a great deal of time over, and one thing that we needed to do was to create an analogy that we could use, and we used the analogy of blankets because it's something that everyone everywhere in the world understands what blankets are, and different designs, different weaving methods, different fabrics and materials and so forth representing genomic variation, so that was a very helpful analogy. And everybody liked it, and I think everybody got it.

Jenny Morgan: I would say that when we're considering knowledge translation and health literacy, we did spend time as a team coming together, and we identified a few ideas of analogies that we could use, and one of our team members proposed the blanket, and that resonated with everybody, and we had further discussions on that and saw how it was very specific where indigenous populations would understand but even as Patricia said, almost anybody would understand that analogy, so we did get a lot of positive feedback where participants felt we were able to translate a very complex medical information in a very informative way that they were able to understand what the challenge and issues were.

Patricia Birch: Regarding engagement, I should also say that now that the journal article is available on paper, I sent the URL to all the participants, and I've already had five emails back from people, so recall that we only had 30 participants, and not everybody had email. People were obviously engaged and are still engaged, and we've had some very positive comments, so I think that also speaks to the effectiveness of the method that we used.

Kate Wilson: I think so, and I think that you both have shared some extremely helpful insights for others who might be looking to do something similar who are interested in research surrounding topic with indigenous Canadians but also thinking about some of the other populations, ethnic groups that we work with as well over in the US. Are there any other resources that you can recommend to those who are interested in these types of studies or interested in maybe pursuing their own types of research?

Patricia Birch: I think I would definitely recommend them to take the time to watch the 25-minute video because I think that obviously they wouldn't be doing the same thing, but it's a good model, and likewise, the community-based participatory research design is something that's a framework that would be worth learning about I think.

Jenny Morgan: And I would say that a lot of the approaches that we use for indigenous populations, especially when we consider the history of colonization, that any researcher doing research with this population to consider different sources of information from scholars or work that looks more specifically at cultural safety for indigenous peoples. And then also looking at the context of the work that it's based in, so here in Canada, we encourage everybody to read the Truth and Reconciliation Commission reports and calls to action as well as the United Nations declaration on the rights of indigenous peoples and just to be more informed of some of the history that's happened with indigenous people so when you're moving forward with research, you're not at risk of replicating some of those same mistakes of colonial approaches or processes that often still happens.

Kate Wilson: I think that's very helpful and something good to keep in mind. Like you said, I think as genetic counselors, as a community, it's something that we want to be mindful of, and we know about the turbulent past with research in indigenous

communities, so I appreciate you both sharing your study but also your experiences with us. Is there anything else that you would like to tell either about your study, the work that you do?

Patricia Birch: I guess there's one thing that I'd like to mention, and that is that there is a much, much larger study that will be sort of moving forward from this, and it's called Silent Genomes, and it's a Genome Canada-funded study, and if anybody's interested in that, I think it would be a good idea to just Google Silent Genomes, and there's a website, and you can see where we're going with that. It's not our group, but it's related individuals.

Kate Wilson: And Jenny, is there anything else that you wanted to add?

Jenny Morgan: Just to summarize what I often remind those around me who are working with, always remember that the process is as important as the final results or the outcome, so just keeping that in mind and just being very aware of the process that's used. If you look at one researcher, his book is called Research Is Ceremony, so using that analogy of decolonizing ourselves as we move through the processes of working with indigenous communities is something just to always be aware of.

Kate Wilson: And thank you for letting us know about that. Interested in learning more about Jenny and Patricia's work? Visit our show notes for the link to their YouTube video. That concludes today's episode of the NSGC podcast series. Thank you for joining us to learn about the importance of understanding and exploring diverse patient populations. For more information on each group's research, visit the Journal of Genetic Counseling under the publication's tab on the NSGC website. This recording is produced by the National Society of Genetic Counselors. I'm your host, Kate Wilson. We'll see you next time.