

**Meaning of Family History**  
*Angela Trepanier, M.S.*

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DR. TUCKSON: We are in the midst of this discussion. I think we have some very provocative issues out here to grapple with, so these next presentations are going to be key.

I want you all to make sure you are locking down on, again, what are the determinant issues in your mind that would cause you to lean one way or another around creating another subcommittee of the Committee and all the work that it entails. If you were going to do that, what would be the charge to that committee. So keep thinking as you are asking these wonderful questions.

So, are you going to tell us about Angela?

DR. McGRATH: Thank you. This is Angela Trepanier from the American Board of Genetic Counseling. Thanks for coming.

MS. TREPANIER: Hi. Actually, I'm from the National Society of Genetic Counselors. Sorry. There is an American Board of Genetic Counseling, but I'm the president-elect of the National Society of Genetic Counselors. I'm also a program director of genetic counseling graduate programs.

I'm here today to talk to you about genetic counselors' roles in promoting the integration of genetic services into health care. I have framed my comments on the basis of a review of Chapter 6 of the SACGHS Genetic Testing Oversight Report, which was Chapter 5 when I read it the first time, so I was a little confused. But same chapter, different number. Then, also, based on the information that was provided from the 2004 educational resolution.

In brainstorming with colleagues, what we identified were three issues that are really critical to integrating genetics into primary care. The first issue, I think, is that we all need to be on the same page with regard to definitions of commonly used terms like "family history" and "genetic counseling." What do those mean in the different contexts of different healthcare providers.

Second, I just think we all need to be cognizant, which I have already heard, that genomics and genetics and health care is an expanding, moving target. What you are thinking about today in terms of integrating genetics into health care could be completely different than what you are talking about in three years. So I thought it was really timely to revisit the SACGHS 2004 resolution because a lot has changed in the last three years.

Then, finally, I think that there is not a "one size fits all" solution for any health care profession. When we are thinking about solutions for integrating genetic services into care, we have to think about practice factors that are going to make the solutions that work different for different providers.

That is how I'm going to frame my comments, and then I'm going to talk to you about what we are doing as genetic counselors to try to address some of these issues.

First of all, I wanted to describe what is the scope of genetic counseling services. This is a comprehensive definition which is based on the NSGC's recently developed scope of practice, the American Board of Genetic Counselors' practice-based competencies, which guide training programs, and then NSGC's definition of "genetic counseling."

I'm not going to read through this list, but this is what we consider as genetic counselors as comprehensive genetic counseling. I'm not advocating that this is everything that you have to do for every patient who has some sort of genetic indication. Obviously, that is not the case. As somebody alluded to earlier, this is not in the best interest of all patients for all indications.

But when we are talking about genetic counseling and incorporating it into health care, I think we have to be very clear about what we are asking people to do. If it is this comprehensive list, then perhaps it is time to refer to a genetics professional. But if it is only pieces of this, then perhaps this could be very easily incorporated reasonably into primary care.

I also want to focus on the family history component of genetic counseling services. When you talk about family history, even when you are talking about the three- to four-generation pedigree, that means different things to different people.

In genetic counseling, what that means to us -- and I'm going to read to you from our practice competencies -- is that we can elicit an appropriate and inclusive family history that we can construct the pedigree using the appropriate symbols, that we can structure questions for individual cases and probable diagnosis. That means asking targeted questions based on a deep understanding of the genetics and the natural history and the features of the conditions that we are trying to rule in or rule out.

That we use interview skills to facilitate recall of symptoms that might not be things that people automatically tell you up front, and that we also pursue pertinent history with regard to how the family has been coping with the condition in question.

That is what we are talking about when we are talking about comprehensive family history. Once again, I don't think this is what we always mean when we are talking about incorporating family history into primary care. In some cases, yes, and in those cases, maybe then you need to think about referring to a genetics professional. But in other cases, no. So we need to clearly define what we are asking people to do.

The next point was really what is the role of healthcare professionals without specialty training in genetics. The role is a moving target, as I mentioned. In the Oversight Report, Hayflick's definition was used, and that definition, which is listed on the right side of the screen, I think is still probably appropriate for many genetic scenarios, many of these single-gene conditions. It may be applicable to other scenarios in the future, too; I think that remains to be seen.

But in 2007, obviously there is a need for roles that extend beyond just identification and referral. By sheer virtue of how much genetic information is available and how much can potentially be used, clearly there are not enough genetics healthcare professionals to address all those potential uses. So we have to figure out a way to incorporate genetics into primary care service just because there is a need to do so.

The gap that has been identified in the Oversight Report and that I have heard from other people, too, is when do you refer to genetics and when do you actually manage in primary care. One of the gaps is just evidence-based medicine, the data that supports when an issue needs to be handled by somebody specifically trained in genetics and when it doesn't.

The third set of factors that I think are really critical in terms of incorporating genetics into primary care are what I call kind of practice factors. Some of those factors have to do with just the disease itself. I think you can develop some practice guidelines based on what you know

about the complexities of the disease genetics, the complexities of testing, the complexities of the management, the potential psychosocial impact, the complexity of decision-making involved in dealing with the disease risk or the condition risk, and then the quality of available data, the degree of ambiguity.

There are also other factors that are important in trying to answer this question, and one of them is provider factors. It is whether or not there are competencies that have been developed for providers giving them guidance in terms of what they should be able to do and potentially not be able to do.

It is adherence to those guidelines and those competencies. As you begin to incorporate genetics into primary care and into curricula for medical students, there are going to be some people who have been out in the field who have not had the benefit of getting that information. So there are going to be those differences in the provider's ability to provide genetic services.

Then there is also practice setting and time constraints. No matter how good your guidelines might be and how clear it might be what primary care physicians should do, there are going to be some practice settings that don't lend themselves to incorporating some of this information into the practice. Then there is also the interest of the provider.

The third factor is really the availability of genetic services in the community and the community's willingness to utilize these services -- in some communities there may be a hesitancy to seek genetic services -- the community's access to information about genetic risk, and insurance reimbursement.

All of those factors also need to be taken into consideration when you are figuring out the best model for integrating genetics into primary care.

I wanted to give you an example. This is a case of cystic fibrosis genetic testing, so carrier screening. There really are in this case two indications for CF carrier screening. There is just population screening. In the Oversight Report, it gave some history about when population screening was recommended for everybody who was pregnant or planning a pregnancy. Then there is screening for cystic fibrosis carrier status related to family history.

The purpose of the slide is to identify where the roles overlap when this is provided in primary care in terms of population screening and when there is some difference in what the roles might be when it is because of a family history.

In both cases, you want to identify and introduce the risk. You want to contract. What does the patient want to know, what information do they want to seek. In the population screening, you need to take a family history. You want to make sure there is nobody with cystic fibrosis in the family history. But it is probably more limited.

On the family history side, you want to take that family history, which is more comprehensive, and you are going to want to know what the family's experience was with cystic fibrosis.

You are going to do limited risk assessment on the population screening side. It is going to be more detailed because you have a family history. You are going to do limited education on the population screening side, more extensive potentially on the family history side. Both are going to require informed consent. More likely, psychosocial counseling will be required if somebody has a family history, and then you are both going to do follow-up.

Where they differ in terms of genetic testing, even though it is the same genetic test potentially that is going to be involved, because of the different types of mutations that can occur when somebody has cystic fibrosis, you are going to need some genetics expertise to tease out what is the best test of the different types of CF carrier tests that are available for this family.

What is required for primary care providers to effectively provide genetic counseling for cystic fibrosis when it is population carrier screening is really to know to whom to offer the test and how to take the family history, basic information about the symptoms, patients' baseline risk, how to accurately interpret test results, how to refer for genetic services when needed, and then the importance of complying with laboratories' requests for patient information, which plays into interpretation of the results.

Then you also need laboratories that provide interpretable test results that people can read and use and that have professionals available to answer those questions, and then accessible educational resources. Genetic counselors can help with all of these factors.

Because we believe that genetic counselors are really integral in not only providing genetic services but also in advocating about genetic services, we have developed this kind of two-pronged approach to integrating genetics into primary care.

The first prong is really training more genetic counselors and making sure that genetic counselors are adequately prepared for changes in genetic medicine. The second approach is to plan educational programs, conduct presentations for practicing professionals, and everything related to educating healthcare providers and the public about how to integrate services into their practice.

Then, just a little bit of data. This shows the increase in the work force of genetic counselors, and this is people entering into genetic counseling graduate programs by year. There is a little dip, but that is related to the fact that this data is contributed voluntarily through the American Genetic Counseling Program Directors Association.

Basically, the bottom line is that there has been a slow increase in the number of trainees entering in programs, up to 205 in 2007, and the number of programs has increased from 18 to 31 since the inception of the American Board of Genetic Counseling in 1993. So there has been progress in that respect.

The number of certified genetic counselors has increased from 495 to 2,437 since 1993. Since the 2004 educational resolution, almost 1,100 have been certified.

ABGC, which is the certification board, had its first cycle of recertification in 2006, and 316 people recertified and an additional 122 voluntarily recertified. Now the certification exam is on a two-year cycle, so we are definitely making improvements in getting genetic counselors certified.

We have been working towards promoting cultural diversity in the profession. There was a program directors retreat that was held with midwest program directors to identify some of the factors involved in why there is not more diversity in the genetic counseling profession. What that resulted in is an improved understanding of what the barriers were and some action points. One of the action points was to develop this brochure that you see on the right-hand side.

Other things in terms of improving our education and training. The American Board of Genetic Counseling is conducting a practice analysis in 2008, and that will help them validate the certification exam and make it an even stronger exam.

State licensure. We have had seven states that have passed licensure bills total. We have five that have introduced bills, and 13 have begun the process. So we are working towards more professional recognition and protecting the public.

Finally, the NSGC is pursuing federal recognition of genetic counselors by drafting legislation that, if passed, would amend the Social Security Act so that CMS recognizes us as healthcare professionals.

I would like to also talk briefly about how we are educating other people. This is data from our Professional Status Survey that show what genetic counselors are doing to educate others. I won't read through all these numbers, but the bottom line is that a majority of genetic counselors are involved in educating all other types of healthcare trainees and professionals.

The types of activities they do are very diverse, and they include speaking to lay and community groups, organizing conferences, coordinating or serving on advisory boards, developing genetics curricula, serving on committees, and developing brochures, pamphlets and videos.

Here are some more efforts that you have in your handouts. I just wanted to highlight that some are Web-based, some are in-person educational comprehensive programs. Many are guest lectures. Some are educational material. So, multiple different models.

Then, NSGC in particular has done a number of efforts to train non-genetics healthcare professionals. We have developed a speakers bureau. We have a whole issue of the Journal of Genetic Counseling that is devoted to genetics education. We have representation on key groups that are looking at how to incorporate genetics into health care, and a whole list of other activities.

So the bottom line, I think, is that genetic counselors have the training, the expertise, the motivation, the expertise, and the track record to be key providers of genetics education. For that reason, I think that it is important that not only do we continue our efforts to train other individuals to incorporate genetics into their practice, meeting them where they need to be met. I think that was an important point that Dr. Khan made.

When you are working with primary care professionals, you have to see what they want and what their perspective is and not come to them from your perspective. If you train more genetic counselors, then not only are you increasing the genetics work force but you are potentially also increasing the number of non-genetics professionals who will get training in genetics.

With that in mind, my recommendation for the SACGHS resolution as an additional step is that not only do we need to support training of other healthcare professionals in genetics but we also need to promote and support initiatives to increase the genetics professional work force, its diversity, and cultural competence.

This can be achieved by supporting the development of genetic counseling programs, providing scholarships to support matriculating students who are from underrepresented minority applicants, and supporting initiatives to increase the number of M.D. geneticists, laboratory geneticists, and genetic nurses.

SACGHS Meeting Transcript  
November 20, 2007

I also have some comments with regard to the Oversight Report that NSGC will submit formally in writing. Because of time constraints, I'm not going to go into those right now. But thank you for your attention.