Role of Professional Societies
Melissa Fries, M.D.

DR. McGRATH: Our next speaker is Melissa Fries with the American Academy of Medical Genetics.

DR. FRIES: Good morning. I'm Melissa Fries, and I'm going to speak to you from two roles, one as a practicing medical geneticist, and two, from my role as the chair of the Education Committee for the American College of Medical Genetics. In this, I hope to bring out a little bit of the discussion of some of the ongoing issues relative to the education of medical genetics, what practice actually is like for someone in medical genetics, and what are the roles of our professional societies to assist us in this.

The education right now for a medical geneticist is a residency, formerly a fellowship, prior to 1992. At the moment it is a residency program for which there are 48 ACGME programs in medical genetics. It is a two-year program. Some institutions have three-year programs with other requirements. There is a prerequisite of two prior years of some initial residency training such as pediatrics or OB/GYN or internal medicine.

There are also five-year combined pediatrics and genetics programs and internal medicine and genetics programs. There are several fellowship programs, such as that for maternal/fetal medicine and genetics, which is a four-year fellowship, and a molecular-genetic pathology program, which is a one-year program. All of these residencies are ones that can be entered into after you leave medical school.

The medical genetics residencies [have] 196 positions. I went through the whole listing and counted them. Forty-seven percent of them are filled. That is a staggeringly low fill rate. For anyone who works in ACGME, you recognize that in most places, like family practice, 93 percent of those positions are filled. We are looking at some of our programs where there are four positions, of which there is one fellow or one resident. The fear of all of the programs is that maybe this year we won't have any applicants.

Clearly, this is a huge issue. The positions are there, the programs are in place, but they are not being picked up by medical students.

This has been the subject of a considerable amount of research. The Banbury Summit report in 2005 -- actually, the summit took place in 2004 -- included representatives of many of these major genetic professional organizations both from the United States and Canada. Canada is a key player in much of this and actually has many of their own medical genetics residencies.

The fact that the programs have not filled means that there is a declining number of people that are going to be available to meet the oncoming role. Many people in genetics look like me. This is not artificial hair. So we need new people to come into our program in order to actually take our places. The whole job in medicine is to train your replacement. That is not going to happen if we don't increase this.

The Banbury Summit recommendations at that time worked very hard to try and reach some consensus on increasing recruitment. They wanted to position medical genetics as ideal for students who were seeking an academic career. Clearly, genetics has to go hand in hand with ongoing research and ongoing practice development, so if you have someone who is really interested in that, that is the person that maybe should consider medical genetics.

Their goal also was to seek NIH funding for centers of excellence and to enhance the visibility of medical genetics by working directly with resident and medical student advisory groups.

There was also recognition of the need to strengthen some of the core training issues as well as partner with other medical specialties and work with some of these joint specialty fellowships. That is where the LFM Genetics Fellowship actually came from.

These have not been idly dismissed, and the continuing process recognizes through Banbury II, which was just recently held -- the report is in press -- that we have to redefine some of our training. We actually plan a Banbury III.

I think one of the things that you want to bring up with the recognition for this is that medical geneticists have a unique role in caring for someone who has a genetic condition. They may actually be the ideal person to be "the medical home" for that person. As you move from your diagnosis as an infant into your role as a teenager, into your role as an adult, you may find that the medical geneticist is one of the key people to actually be able to do that. That requires a change in the training. If you are trained largely to think of things pediatrically, you are not going to be able to actually follow through then into their role as an adult.

Speaking in my own practice, I come from a circumstance where I spent 26 years in the Air Force, where I was an OB/GYN and a practicing geneticist. Most of my role there was in prenatal diagnosis and genetic consultation.

I then moved to a practice in inner city D.C. at a largely academic but very busy inner city hospital. I'm the only geneticist. I'm called the director of genetics and fetal medicine. I am in charge of myself.

[Laughter.]

DR. FRIES: Which is really helpful. But this gives you a sense that even for those places that have medical geneticists, they are rare birds.

What I find is that my practice is guided by these three Rs. The first one is recognition by other professionals. You would think there are so few of us that maybe the hospital, by just mere fact of hiring me, would make some effort to market me. It hasn't happened yet. So marketing and advertising what this person does is, I think, a key function and one of the things that could be done very well.

I was working on an initiative in my own institution for this intranet curbside consultation. If you have that in your own intranet system, just as Beth was commenting, you can then click on that and then, one of these hours when I'm not doing other stuff, I will try to get back to you and tell you if it is one of those that we could work on.

The lack of recognition then leads to my second R, which is the referral process. Many providers, even in fields where you know that there are genetic issues, don't feel a need to refer. They often feel that they can handle those genetic services just as well and it is probably not going to be a beneficial role for the patient. So the referrals are struggles.

And then the final R. I know we have heard a lot about reimbursement, but I have to tell you, the issue of reimbursement in my own institution has created a two-tiered system of genetics because most of the patients are Medicaid. Medicaid patients cannot get a genetic test paid for that is out

of state. So if I want to get one of my Medicaid patients tested say for BRCA1 or 2, she either has to pay for it herself or she has to go through Myriad's need program, or hopefully will be able to pay for it through a grant.

But what happens is that then you get an insurance quandary so that you are tiered for that, and patients may not even be referred because the issue of the reimbursement is such a problematic one.

One of the other issues that you find in practicing in a diversity of medical settings is that there is this ongoing pattern for use of family history. Family history we want to incorporate into all medical fields, but even for experienced genetic providers who do medical family history-taking or genetic family history-taking every day, across demographics this is very difficult. The socioeconomic issues and the cultural issues are enormous.

Immigrant populations may have minimal information as well as problems with literacy and language. We are not just talking about Spanish. I spend a third of my time speaking Spanish to my patients. I have gotten a lot better. But at the same time, my patients may not read Spanish. How are they going to deal with that. How do we deal with those literacy issues.

They may lack information on their parentage. Things that we think, okay, mother, father, you are going to know this, that is not always the case.

The medical issues in the family may not either be discussed, because they may be taboos, or they may be in some ways certainly unknown. So I think that one of our key areas in this is to focus on the development of tools and education across these demographics of language, culture, and literacy. This has to be a key point for integrating this truly into practice.

I cribbed this from Dr. Charles Epstein's article from 2005 about medical genetics, but this basically shows the pedigree of the institutions that are here to help us as professional organizations. You can see that the parent organization, the American Society of Human Genetics, has been around for over 50 years and has given birth to quite a very few children. Actually, they were born quite late in life, although they seem to be still fairly robust.

In 1991, we had the American College of Medical Genetics. In 1980, we had the American Board of Medical Genetics. American Board married late and now is part of the American Board of Medical Specialties, and we have given rise to our sole child right here, the RRC for Genetics in the American Group ACGME. So this is our group of professional medical organizations that are associated with the practice of genetics.

They have all different roles. I would say that one of the key overwhelming roles of all of them is the recognition of the importance of education. I think perhaps in no other field does education play such a huge role. Any genetics interview, any genetics time, is education. It is education for the patient, but in many ways you are educating whoever is around you: your nurse, your genetic counselor, your high school student who is watching over your shoulder to model behaviors.

Geneticists educate as part of their life's blood. So all of our professional organizations recognize this. The American College of Medical Genetics has as its goal the education, resources, and voice for the medical genetics profession, to make genetics services available to and improve the health of the public in general.

The American Society of Human Genetics is a very, very broad organization, but the Information and Education Committee's goal is to identify and promote educational opportunities to increase the understanding of human genetics in North America. They have several specific focuses for that.

Our American Board of Medical Genetics is our certifying organization. This is how to keep us current and how to maintain our certification both for ourselves and for our training programs.

ACMG has been a powerhouse in working these educational initiatives. We discussed the importance of making genetics part of board examinations. The American College of Medical Genetics has sent several taskforces, at least four times in the past 12 years, in conjunction with the American Society of Human Genetics and the Professors of Human Genetics, to review the questions on USMLE Part 1, 2, and 3.

In looking at them, we found that there are definitely improvements, and this is from Darrell Waggoner's presentation which you just commented on. My [apologies] to Darrell Waggonner because I misspelled things here. I apologize.

But there is definitely an improvement in the incorporation of these basic science questions. There is an increase in the part 2 and Part 3, but the irony is that very often when they give a clinical scenario, family history is not part of it. The patient is presented. A 57-year-old man presents with chest pain and a cough. You don't know that he has a family history of hypercholesterolemia and that he has a family history of diabetes. Any of those other family histories are just not given. So clearly, still, efforts need to go on.

There is definitely some improvement. We hope that this will be expanded with use of virtual patients and clinical scenarios.

ACMGE is also working on the exposures of general clinical genetics with video teleconferences. I would invite you to go to this website, Neurofibromatosis, UnderstandingNF1.org, where there is Bruce Korf interviewing someone who has neurofibromatosis. The intent is to develop about 10 of these video telecasts so that people can have an idea of what geneticists actually do and provide models for those who want to actually look at what this role would be. What is your job going to be like.

ACMGE is also involved in looking at the residency curriculums. We talked about this. This is a collaborative effort to promote the idea of our medical geneticists as the medical home for lifetime care for some of those people with congenital anomalies and genetic conditions.

Another key point we have also addressed is the idea of expanding point-of-care reference systems. ACMGE has developed things called ACT sheets. The ACT sheets are in response to the expanded newborn screening programs where there are at least 29 different things tested for, of which they may come back with positive findings leading many people in the field to both weep, tear their hair, and panic. The ACT sheets are very accessible and very knowledgeable.

One of the interesting and very important issues right now is to incorporate these directly into our electronic medical record systems so that there is an automatic pop-up for them. These protocols are going to be similar models for other activities, such as those on cystic fibrosis, Fragile X, hemoglobinopathies, and then could also be involved with how we work this patient up. What would be ways that could be guided for development of studies on mental retardation or

developmental delay, and how do you work towards this. This is actions of the American College.

American Society of Human Genetics has focused on a different aspect, not so much the medical but on the overall understanding of genetics in general. Charlie Epstein, in his presidential address, emphasized that one of the key things that the public has is a fear of genetics, a fear that genetics is going to somehow make a superhuman person, someone will be basically made and we will no longer be able to have our wonderful diversity, that there will be priorities of what is good and what is bad. This is a chronic fear of the public.

The American Society of Human Genetics has worked on this in their expansion of programs K through 12, and actually K through 16. There are developments of programs. There is a program called GenEdNet.org, a little bit hard to say but a very worthwhile program. There is a database of genetic standards for education at the K through 12 level across all states. So if you want to know how to teach genetics to a kid in kindergarten, you go to GenEdNet.org and you can find out for your state what they will do. It is a wonderful program for all of that.

There are numerous other initiatives that have been involved: DNA Day, essay contests. There is a program called Genetics Education and Outreach, and there is a grant right now that the American Society of Human Genetics has of pairing a geneticist with an educator for training and education.

Clearly, working through the schools is the way to incorporate basic genetic knowledge because your kid is going to be the one taking that piece of paper back to the family and saying, "I want to know what grandma had and what grandpa had." The child is going to be the mover in that particular field.

ASHG also runs a wonderful undergraduate workshop with every meeting that they have, where they are going to be incorporating students and undergraduate educators as well as high school educators from the community in which their meetings are held. There is a key emphasis on education as part of your role as a geneticist.

Finally, the American Board of Medical Genetics is very active in our maintenance of certification, which all of us must meet as physicians. One of the points I would like to emphasize is in our Part 4, where we want to improve our practice models. We will write genetics modules for that that can be translated to other specialties for their utilization in that particular area of training.

I would like to conclude with some of my own thoughts about recommendations for this. There is clearly an improving trend in some points of medical genetics, but it is not enough. We need research on why people make their choices for residencies. A lot of it, I believe, is related to the fact that they don't know anything about what medical genetics does or is.

I would also like to suggest that maybe there is some role for a sponsorship program. If we recognize that medical genetics is a key profession that needs providers, maybe there is a role for a sponsorship program much like we sponsor those who serve in inner cities or rural communities after their training.

I think we all have to recognize that if we are at an academic center our practice patterns are going to reflect some of our initial specialty training. Judith will address a little bit more of that.

But I think that we need to recognize within our institutions some of that. We also need to work with this issue of reimbursement.

Finally, I think that all of our professional societies work for education, but it is a work in progress. Education is not enough, as we have spoken before. You have to put it in practice, and you have to develop a competency to reflect that you actually can use that.

Thank you. I welcome any questions.