

## **MCH/CSHCN Director Webcast**

May 12, 2005

CHRIS DeGRAW: Good afternoon and welcome to mchcom.com webcast coming to you from Rockville, Maryland. This is an Internet webcast for state maternal and child health directors. I'm Chris DeGraw from the Division of research and education. Dr. van Dyck had a continuity of operations drill this afternoon and wasn't able to make it. We have an interesting program today featuring Dr. Alan Guttmacher talking about the U.S. Surgeon General's family history initiative. I want to review technical information about the webcast. In response to your suggestions the speaker's Power Point presentation and accompanying article are now available on the mchcom.com so you can download the slides and handouts before the webcast. Slides will appear in the central window and should advance automatically.

The slide changes are synchronized with the speaker's presentation. You do not need to do anything to advance the slides. You may need to adjust the timing of the slide changes to match the audio by using the slide delay control at the top of the messaging window. We encourage you to ask the speakers questions at any time during the presentation. Simply type your questions in the white message window on the right of the interface, select question for speaker from the dropdown menu and hit send. Please include your state or organization in your message so we know where you're participating from. The questions will be relayed onto the speakers periodically throughout the webcast. If you don't have the opportunity to respond -- if we don't have the opportunity to respond to your

question during the webcast we'll email you afterwards. We encourage you to submit questions at any time during the webcast.

On the left of the interface is the video window. You can adjust the volume of the audio using the volume control slider which you can access by clicking on the loudspeaker icon. Those of you who selected accessibility features when you registered will see text captioning underneath the video window. At the end of the webcast the interface will close automatically and you'll have the opportunity to fill out an online evaluation. Please take a couple of minutes to do so. Your responses will help us plan future webcasts in this series and improve our technical support. At this point I would like to turn the program over to Dr. Michele Puryear. She's from our division with children with special healthcare needs.

DR. MICHELE PURYEAR: Good afternoon. A number of federal agencies have worked together over the past year to design and implement the U.S. Surgeon General's family history initiative. To help promotion and disease prevention. This presentation will describe that initiative. Dr. Guttmacher is a graduate of Harvard college and medical college. He completed his residency in pediatrics and fellowship in genetics at Boston hospital and Harvard. A member of the Institute of Medicine. A fellow of the American Academy of pediatrics and the college of genetics. He is the deputy director of the national and human genome institute of the national institutes of health and director of the Office of policy and communications at the genome institute.

Before I turn you over to Dr. Guttmacher, I want to talk about why the Maternal and Child Health Bureau feels family history is important. The events and experiences of your everyday lives are part of history, part of your personal history and part of your family history. History books are record books of official events. They often tell only the stories of presidents, kings, army generals and famous scientists, writers, artists and inventors. They usually don't tell the stories of ordinary people. When you learn the life stories or oral history of your parents and grandparents, it can help you understand history better. The stories they tell you about their own lives can also help you understand your own history better. Their stories are your heritage. You inherit many things from your parents and grandparents. They transmit a cultural heritage and values, through recipes, certain foods, family customs, religious practices, the love of stories, songs, music, favorite dances or family heirlooms such as a quilt. These traditions contain valuable knowledge not found in books.

You also inherit physical qualities. The color of your eyes and hair, body and size from your parents and grandparents. These physical qualities are passed down to you through your genes transferred to you from your parents when you are first conceived. Genes can also pass on the possibility of developing certain diseases or the possibility of having excellent health. Your personal health is influenced by that family history because family members usually share genes as well as physical environment. Like the places where they live or work and lifestyle, the kinds of foods they eat, the kinds of exercise they get, whether they smoke, learning your family history can help you understand your own health or illnesses. If members of your family share certain health problems you might be at risk

to develop those in the future even though you're healthy now. If family members have found ways to stay healthy perhaps you can learn something from them, too.

We feel that your family history is one of the most important pieces of information you have about your personal health. Some consider family history the first genetic test. It's not found in any book, only found in the memories and experiences of your relatives. When you ask family members to tell you stories about their past, the places where they have lived or the occupations that they have had, ask them questions about their health, too. If you find out that they have had diseases such as asthma and heart disease, diabetes, certain cancers, you can use that information to help yourself and other members of your family. When you explore your family history by asking a relative to share their memories with you, you'll hear some great stories. You will also learn about information about the health of your relatives that can help you make good choices about your own health. Your family history is rich; you don't want it to be lost. Your family history is very important to your health and learning about it, writing it down, passing it on will be one of the most important things you can do for your own health and the health of your children. So I would like to turn direct to the presentation and turn it over to Dr. Guttmacher.

DR. ALAN GUTTMACHER: Thank you very much, Michele. Thank you and thank you, Chris, as well for the introduction to this. It's a real pleasure to be here to talk with you about what I think is a very important initiative and talk with an important audience for it. I think that the programs that you lead could benefit from using this but I think the initiative

could benefit from your active participation and I'll talk with you a bit about that as we go along. It's a wonderful opportunity. I hope for us to have some discussion at the end about this. I have to apologize to you for one piece of my own family history before we begin and that is I unfortunately have inherited genes from both sides of my family and it has made me so bald that the reflection you're getting as you watch this, the brightness at the top there is not much we can do about that. You're the prisoners of my own genetic makeup. I don't have to worry about it but you who are watching will have to pay the price.

What will we talk about today? We're going to talk briefly about the past use of the family history and then a little bit more so fairly briefly about the current use of the family history. Then spend our time talking about what the U.S. Surgeon General's Family History Initiative is and what it's going to be used in the initiative but other ways to use the family history as we go forward in healthcare. So the past use of the family history is really not a medical use primarily but a genetic one. The way it got started is what I think of the begat age of the family history. This is our lesson for the day from the book of the Bible, Matthew 1:2-16. And basically it's the begat age of the family history. It begins Abraham begat Isaac and Jacob and Judas and his brethren. At the end it goes to the birth of Jesus. It traces Jesus as a direct descendant of Abraham and other examples of this historic use of family history.

If you ask most people in our society who have ever used family history before, what have you done with it, it's been about genealogy. It's not directly a medical use but it does, in

fact, I think, in some ways make medical use more available to us because people are used to this idea of family history. They're used to it in a different kind of setting by going to the family Bible and seeing what is recorded in there or other kinds of stories that have been handed down over the dinner table to people constructing an official family history or an imaginary one in their heads. As we move forward with this.

The next slide, please, we're really talking about the family health history and I would like to divide that into two. One is what we're doing today and what we've done for some years in healthcare, which is to use the family history in many ways as a surrogate for the individual's genetic makeup. We're not yet at a point where we can easily, effectively, inexpensively really understand what someone's individual genetic makeup is. And our quickest approximation of that, it's the first order approximation. It's to look at the family history to give us some idea what the individual genetic makeup is. As we look forward in the future, what we'll do is use family history in conjunction with much more sophisticated, much more widely available, much more widely effective and useful genetic testing and combine the family history with our knowledge of somebody's individual genetic makeup to really predict individual disease risk, to understand prognosis for individuals who do develop disease, and actually even to think about individualizing therapies for various diseases.

Now the next slide makes the point that we're in the genome era now. Two points here. One is that we're in the genome era. Where we have the human genome available to us. We're in a very small period where we're going to have the D.N.A. sequence in hand to

being able to figure out what the sequence means, understanding the genes that are hidden in it and very importantly understanding how those genes translate to health and disease in general in terms of understanding the biology of disease and of health. But also understanding the genetic makeup of the individual and what role that plays in health. The second point is that we're in the genome era but not the post-genome era. You're talking about people saying we're in the post-genome era. It was finished on April 14, 2003. Until that point in time for all of humanity's history we clearly have been in the pre-genome era.

For now the post-genome era it was suggest that the genome era was April 14, 2003. If you slept late you missed the era entirely but you didn't. It's all right if you slept late that day. We're just entering that period in time where we're really beginning to use this knowledge of the genome to make an impact on health. Eventually folks will be in the post-genome era. I for one will be well retired by then. Most of you will be as well. We look forward to that but we have important work to get to that era. So in the age of genomics why would we use family history? One reason why we would do that, the basic reason, is that diseases are due to the interaction of multiple genes and environmental factors. Most of these, some people would say all diseases are due to interactions of multiple genes and multiple environmental factors.

As the next slide shows, if we look at the ten leading causes of death, you can do instead of mortality if we looked at morbidity it would be a similar kind of story. We look for the ten leading causes of death. These are the last year which we have data available from the

C.D.C. 2001. It doesn't change much from year to year. It doesn't change that much from one developed country to another, in fact. If you look at the ten leading causes of disease, what they have in common from a genetics point of view is that for years we've been able to say for almost every single one of them that family history matters. That if you have a family history of heart disease you're at increased risk for heart disease. We haven't been able to say very much beyond that. We've been able to say you have an increased risk and probably should worry more than other people. But we haven't been able to guide prevention or treatment or prognostic kind of strategies based on that.

What we also have in common for these diseases is we're in this short period of time where we're beginning to understand the individual genetic factors that have been responsible for that disease risk that's been shared in families. Specific individuals have increased risk for heart disease and why and begin to design individualized prevention and treatment plans that would make a difference for individual health. And all ten of the things on this list. There is one on this list that if you look at the slides carefully injury that may some people would say would deserve a question mark rather than a check mark next to it. What could appear to be more random than injury? Well, in fact, genetics may play a role there in a couple of ways. One is it's a well-known genetic factor that you don't have to do sophisticated genetic testing to recognize. You can do crude physical diagnosis to identify individuals who have this genetic variant that makes them at increased risk for dying of injury.

I'm sure that many of you watching this, I hate to tell you, but you'll have this very common genetic variant that does make you more likely to die of injuries. It's something known as the Y chromosome. Those individuals who have that genetic variant will be a couple of things. One is they'll be male. The other thing as my wife points out. The Y chromosome made nothing useful for you. It makes you a male and also increases your risk of dying of injury. Those folks in the audience who lack the Y chromosome and are females have a lower risk of dying of injury. That may not be biological or genetic. It may be socio-culture. Even if we forget the Y chromosome it is true for one's risk dying of injury is modified by genetic factors. Not necessarily your risk for getting injury. Once someone gets an injury who dies and who doesn't die? We tend to think of genes as causing disease but they play a role in once one gets a disease is how you act to it. Who has an injury and ends up in the hospital. Lots of variables play a role there. One of the variables we're beginning to have evidence and identify specific factors that play a role is one's genetic makeup. The genetic makeup you have will react to the insult that you've seen and will help dictate how you react to it. So here even in this one genetic factors play a role.

As the next slide emphasizes for none of these is it genes alone. It's genes and environment that add up to equal health and disease. As the next slide makes a point, you can sort of do a geometric kind of proof here or something, after all, family his trees -- history is a record of health and disease. Therefore, just by doing a substitution there it makes the point that Michele was making in the introduction. Family history is a record of shared health and disease which means we share genes and shared environment. It is

not that precise for either but is a good first approximation. It tells us something both about the genes and the other cultural and dietary, etc., elements that are environmental. Non-genetic that we've also shared with other family members.

The next slide goes on to make the point that family history today can change the way we do many population screening kinds of things. I'll get back to this in a minute. But it's an important point I think particularly from a public health perspective. It's important to realize this sort of genetic use of the family history has a role in individual medicine but also has a role in public health practice. That is, it can help us perfect the individualized screening mechanisms we do. I don't have to tell this audience, public health screening measures are the major reason, not the sole one, the major reason why we all live healthier, longer lives than folks did a century ago. Let's admit it. Most public health screening measures, the way we use them today is a pretty blunt instrument. If you're in your 40's, you should have an annual mammogram. We don't know whether that's correct advice. It depends who the woman is. If a woman has certain mutations which puts her to increased risk for breast cancer maybe she should start having annual Mammography in her 30. In terms of saving our public health efforts, in terms of saving dollars and really being much more guided to what an individual should go to benefit themselves, maybe that woman -- some women should not have it until in their 50's. We have good evidence-based medicine to show that knowing family history can influence how we even do population-based screening.

And the next slide makes the point that for people who develop common diseases, that again knowing family history can change the way we do management for people once they became ill. I'll get back to an example of that a little later. Evidence-based medicine to let us know that family history can make a difference in this kind of thing. So do we have any evidence, given all that, that the public cares about family history? We have a couple of pieces of data that suggest the public does care. One from last fall, a parade magazine research America does an annual survey each fall. In 2004 they asked one question to give you a context. They asked people, have you found health information on the Internet to be very or somewhat helpful?

>> Less than 2/3 of people said that was the case. But more importantly to us the next question, the next slide, another question they asked was, do you think that knowledge of family history is either very or somewhat important to your own health? And 96% of individuals said that they thought that knowledge of family history was either somewhat or very important to their health. Which to me was just amazing. I'm not sure that I can come up with any question that I get 96% of Americans to agree on and here 96% said that well, maybe that is some kind of ascertainment bias, something wrong with the survey but we have another piece of data from almost the exact same time.

I think a month earlier which will show in the next slide from a survey that the C.D.C. did of 4,000 people. And they do an annual hope survey as many of you know, this survey asked a similar question, by coincidence. This one, 97% of individuals thought that knowledge of family history was important. So 96 % in one survey, 97% in another seems

it may be real data. A high percentage of the American public thinks it's important. But the next question that was asked may be more telling in some ways. Remember, 96 and 97% think it's important. Another question the C.D.C. Asked was, have you ever actually collected health information from relatives to develop a family health history? Only 30% had. And that is why I think the Surgeon General's family history initiative is so important. We have a public that's aware that family history is important. We don't need to convert them to believe it. They believe it but they haven't had the means to be able to do anything with that belief. So we have an interested public and I think all we need to do is to give them the tools to do something with that interest to benefit their health.

And as the next slide talks about, this is really one of the founding reasons for this public health initiative. Why family history? Several reasons. Again, even though we're going to have lots of new genomic sexy genetic tests in coming years. There is reason to believe that family history will remain relevant for healthcare for many years to come. It is not going to be replaced by genetic testing and that as we reviewed family history does help give very good predictive information in terms of risk for very common diseases including heart disease, colorectal cancer, breast cancer, osteoporosis, diabetes, suicide, other kinds of real public health concerns. At the same time we have people who are unaware of their family's health history. We have many health professionals who underutilize the family history. We have data that demonstrates that and have difficulty in advising patients based on family history how to maintain good health.

The next slide, please. Really as we thought about this there are a couple of things I think that occurred to most of us that were involved in this and challenges we had to confront. One of them is this underestimation by clinicians of the utility of the family history. How do we change that? I think we change it in a number of ways. One of them is it requires better teaching of clinicians of the future, be they physicians or other health providers and also modeling of more effective use of the family history in day-to-day practice whether it be in public health or individual medical care settings. The second challenge is that for people who are busy in terms of healthcare today, as all of us are, is finding the time to obtain, organize and analyze the family history. This is not something one can do in two minutes in a brief clinical visit, how do you find the time to get all this information? Those are the two challenges we were trying to face. The next slide shows, as Michele said before, a number of federal agencies came together to work on the initiative all within the Department of Health and Human Services but a number of different agencies. HRSA has been a major part of this, the N.I.H. has been a part of this, AHRQ has been as well and the C.D.C. But all led by the office of the Surgeon General. The next slide we sort of got together saying what can we do? We came up with the family history initiative in a joint effort that made sense. The reasons for doing this and some of these are things we've already gone over in a way. But there are several goals of this. The first one is to increase the public's awareness of the importance of family history and health. To some degree that 96% 97% statistic -- 100% would be a nice goal but we'll settle for 96% or 97%. We can increase somewhat not who is aware of this but how aware people are of this. Most important we need to give the American public tools to help them gather, understand, evaluate and really utilize their family history to improve health. We need to

increase the awareness of health professionals about the importance of health history and need to give them tools to make their use of the family history something that is more efficient and more effective as well.

The other ideas we would like to increase both genomics and health literacy. The Surgeon General is focused on this. He believes very strongly, it's easy for me to say the genetics is the future of medicine. It's interesting the Surgeon General who many of you know is a trauma surgeon by background is a great believer that genetics is the key to future health prevention strategy. He incorporates talking about genetics in most of the talks he gives across the country about this. We believe we have a window of opportunity take we have a few years until genetics becomes much more broadly utilized in health. That clearly is only a few years away. We need to educate both health providers and the lay public to think genetically. To think more about genetic issues. To think more about the way things are inherited, that kind of thing. The family history is a tool we use today to increase genetic literacy as well as health literacy in general. When we have more genetic tools available in a few years we have both a workforce and consumer force that are ready to use that kind of information. So it's really preparing people for this genomic medicine that we think will soon be an integral part of healthcare.

Next slide, please. So we came up with this thing that's called the U.S. Surgeon General's family history initiative. You can see there is a website. It's [www.hhs.gov/familyhistory](http://www.hhs.gov/familyhistory). If you go there you'll be able to see a lot of what I'm about to talk about. And the focus of this really is a tool. Something called my family health

portrait that allows people to really incorporate and download their health history. Here is a picture of a print version of this. This is available both on the web but also as a print version. It's available both on the web and the print version in both English and Spanish. HRSA has actually spent some of its money in this initiative to print up many copies of this and if people want to know how to obtain it, folks at HRSA can let you know how to obtain print copies of this for folks who come into your clinics but it is also available on the web. The next slide will begin to walk us through this. If you go on the web, you find some welcoming messages that explains and it sort of walks an individual through recording their family history.

The next slide gives one of the pages that asks someone to input their name, their date of birth, say something about themselves, and then the next slide makes clear that you're asked to comment specifically about six common diseases. These six diseases were chosen because these are the six common diseases for, again, we have the best evidence-based medicine that knowing family history makes a difference in individual healthcare. We asked people about history of heart disease and colon cancer, etc. These six kinds of things. There are a couple of dropdown boxes so people can put in other diseases that are of particular importance to them or within their family. I should tell you this tool is pretty good now. We had to get it up for last fall we had to rush it to market so we've been busy since last fall, based upon user response, perfecting it. In two months or so there will be a better version on the web as well. This is a beta in some ways. The new version will allow you to input as many diseases as you want to. The screen that's up there now asks about what other family members do you have, how many brothers and

sisters and uncles and aunts, etc. If you do as the next slide shows, you then are asked to input data about each of these individuals and, for instance, you'll see here this one has indicated that the individual by which that has one of these conditions and if you indicate that, a dropdown box then appears asking them what decade of life did this individual get this disease because there is a different sort of importance for some people develop cancer in the 40's it is more likely to be hereditary than someone getting it in their 60's. It helps guide us in the use of this genetic information.

Next slide, please. The tool then will give you either a visual depiction of the family history of the pedigree or the next slide shows will give it to you in tabular format. The idea that the individual can then print this up, walk into the health provider's office with this information so that the health provider has it. Now, what have we done with this? We've done several things. One is we've moved the collection of the family out of the medical office and into the home. We think that has advantages for several reasons. One is, there is reason to believe this will give you a better family history. Why should that be? That's because if you think about constructing a situation where you're likely to get a lousy family history, do it in a profession -- someone coming to your office as a physician or other healthcare provider not to discuss families but because they have some complaint or they're coming for an annual well visit or something like that. But they aren't coming primarily to talk about family history. Suddenly, tell me about your family history is sprung upon people unprepared for this. They have no records available about their own health history often. Certainly no other records of other history of family members. If that's available at home. You may not be at home immediately but you can gather that

information and most importantly at home you can gather the family history by gathering relatives. Relatives as we all know are the keepers of the family history. The older relatives are the best keeper of this. Don't just ask the older person but ask -- ask what it was that uncle George died of. So by doing that at home the individual has much more recourse to better resources for their family history to make it. The other thing we've done is made the individual -- instead of making the -- when you think in some ways that empowers individuals -- [inaudible] -----

>> This was launched last November with a real focus on Thanksgiving. The idea being that Thanksgiving is a day that many American families gather together and if not Thanksgiving there are other holidays that follow close thereafter where family members tend to gather together. To make the Thanksgiving day the focus of this was the idea to when families gather together get them to talk about family history. There is that down time between the Thanksgiving dinner and the Green Bay Packers game where you need to do something to entertain the guests and stuff. One thing to do which can sometimes be quite entertaining we would advise you watch the amount of liquor served with dinner if you're going to do this as an after dinner event in certain family. We haven't seen any big uptake in family violence related to this but as we all know family history, sometimes different people in the family will have different readings. Family history. It gets back to other issues, it's not just about health but traits or characteristics in various family members. The idea was to focus on Thanksgiving. There were over 1,000 media stories about this last November. So the Word got out there well.

We want to increase the drumbeat this year. We want to make it an annual event. To make Thanksgiving just as early November there is a day which is the great American smokeout day. The Surgeon General is behind the idea of making Thanksgiving the annual American family history day to increase awareness of this, to do different kinds of campaigns. For instance, this year many of you will be aware that the Surgeon General, this is the year of the healthy child. So we would like to link this to the Surgeon General's efforts around 2005 being the year of the healthy child. One of our focuses this year as you move toward Thanksgiving with this.

Next slide, please. So for those of you who can see this slide, this is our Norman Rockwell view of the American family. Now you've done the family history you qualify for the turkey dinner. Next slide, please. Also last fall the Surgeon General, the director of our institute at the N.I.H. The national genome research institute and I tried to write an article to get health professionals to think about this which happened to be Thanksgiving day itself when it appeared. Here is the next question for you. You had to subscribe to the journal to be able to download this. How many times is this downloaded between the time it appeared November 25 and the end of March? So your choices here are 100 times, 500 times, 1,000 times, 5,000, 10,000, 15,000 or 20,000. And unfortunately no matter what you answer for this one, the next slide will show you a wrong because it was 21,000 between the time I originally made the slide and when I went back to check the data a month later. More people had downloaded. Over 21,000 health professionals have downloaded this. It surprised me because I didn't think it was that -- if you have a copy of it, you can judge for yourself. But I think it shows again that health professionals

are aware this is something of potential importance. So that again I think it makes us feel good about the initiative of having a real future. Next slide, please. So we've talked about today, let's talk briefly about the future. What is the future of the use of family history? Well, as I said before, we don't think it is going to replace -- at least not for a long time, with other kinds of genetic testing. It's not that great a surrogate but the best one we have for individual genetic makeup. Once we can really know someone's individual genetic makeup then perhaps family history won't be so important. There will be a number of years before we completely know someone's individual genetic makeup and can really understand that translate into help for them. For a number of years what we think we'll be doing is using a combination of family history with knowledge of individual genetic makeup and testing results to craft individualized disease/risk prognosis, responsive therapy, those kinds of things.

The next slide mentions an example of this. Maybe it doesn't. It could have mentioned.

There is an example of this today that we begin to use these two things together. Let's see the next slide. OK, yes, this is going into the same thing. Let's go to the next one.

This is the idea that even today we're combining the two not very frequently, genetic testing, but this is a paradigm for the future. That is there is a Gene known as BRCA one because it causes mutations and increases a woman's risk for breast cancer and ovarian cancer and a male's risk for prostate cancer. Breast cancer was first associated with it.

An important question is if a woman has a mutation of it what is the chance of getting breast cancer? It isn't 100%. It depends upon her family history, not just her mutation.

For women who have a mutation in BRCA one such woman has a higher risk for breast

cancer than a woman with the mutation with no family history of breast cancer. Family history tells us about other shared genes to act to modify that Gene. It tells us something about shared environment, diet and culture. It may say something about the amount of sun exposure we get. We think that may influence risk for breast cancer. Other things shared within family. For a number of years we think we'll be crafting things about genetics and family history to help individuals. Next slide, please. What might we do to give us better tools for this in the future?

Next slide. Well, one is that we think we need to really better understand the way that these multiple genes and multiple environmental factors interfere with health. This is an article that was written last year in the journal of nature. Which talks about the real need in the U.S. of a very large study of genes, environment and health. A large cohort of 500,000 people we follow over a number of years to tease out this intricate interrelationship of the specific genetic and environmental factors. We need to do this in the not too distant future. We need to effect and make cheaper and more accessible genetic testing. We're working to do that but we have to think about that as not just an issue of genetic testing but a health systems issue. We need to develop techniques for gathering appropriate information about all environmental factors. That's a good example but the sun example is a poor example. It's more than the physical environment. You will know if there are sidewalks. The behavioral environment, social, cultural environment. They can all play a role. We all need very much so to develop and test in the real world ways to collect, analyze, communicate and play all this kind of information. Even when we had elaborate genetic testing, unless we know how to use it in a clinical setting in a public

health and individualized medical health way to change health behaviors and to make a difference in health outcomes, then it isn't worth doing. We need to do a lot of research and working together to figure out how to do those kinds of things.

Next. So someday our hope is that with better genetic testing and ways to employ these things the family history will return to the year of the family history as the begating tool as the genealogy. Next slide. So this is just my reminder to myself to remind you and perhaps to give an example that family history really can be a lifesaver but it does have its limits. I think that we can extend those limits. Let me take a minute of the time we have left to give you an example of this so I don't always do and you'll notice that I did not include in the slides here my personal family history. That's because I'm not sure all my family members would want to be identified by name to all the health departments around the country. You may be looking for them in some of your health departments. Let me tell you about my own experience with this. I don't have a typical experience. I'm a medical geneticist and be sophisticated about my family history. I should have been aware of it. That kind of thing. When I sat down and used this tool what occurred to me.

So I went through. I entered my relatives in the tool. Like any sophisticated medical geneticists called my mother to get help to fill it out. I looked at it and it was interesting. It said a couple of things to me when I did it last fall. There is a lot of heart disease in your family, Alan. Both of my grandfathers died in their early 50's of myocardioinfarction. This is something to have you paid attention. I had a heart attack myself at the age of 49 I had been aware there was a family history of heart disease. What's interesting for me is to

think the family history laid out in front of me looking at that. It's true that I had some years of borderline hypertension before I went on to have heart disease. I used to be significantly overweight. And I wonder if somebody if my primary care providers had sat down and gotten a good family health history, gone over that together I suspect my good internist would have said this borderline hypertension in somebody else we would just let skate. Your excess weight which is behind that, we just would let get by otherwise. You need to pay more careful attention to this. I don't know if that would have prevented my heart attack but I think it might have made a difference.

Certainly in public health ways if you have enough 45-year-olds looking at that and take a more focused interest in healthy behavior to make a difference in their health behavior we may delay and even prevent heart attacks would be a health benefit and save us a lot of money. Here I am a sophisticated person and again you know, a simple tool like this could make a difference. It's interesting my other family members have used the tool, taken it more to heart because of my lesson as well. There is a limit to the health history that I'll mention to you, which is when I filled this out the other thing that jumped out at me my family history is not just designed for a Genetist. I'm the product of an identical twin. My father and uncle are identical twins. Genetically I had two family history for a father that I can take advantage of. My father died of acute leukemia and his brother died of another type of leukemia. I've been aware of that. Put it down on paper. You need to pay attention your father and surrogate father both died of leukemia. Unrelated forms. Though some of us in the family have a theory that my father had it and it wasn't diagnosed that his brother developed CLL later. It's interesting family history. It became

more interesting to me after I filled out the family history when I was diagnosed with having CLL myself. Now, there is an interesting point to me in looking at this professionally that knowing that family history, having filled it out nicely last November and everything, that didn't allow me to ward off leukemia somehow. I think as we look forward to having more sophisticated genetic testing, to have more understanding of factors I would assume for the next generation in my family that that would be a different story. That will be the story much like heart disease today. If someone is aware of that family history and use genetic tests that will be available just as will be much better genetic testing to identify the specific risk factors for heart disease in my family. You see -- using that information can make a difference in people's health. I may not be a routine example but here to give testimony I not only, you know, helped sell this product, I bought the company. I don't know what the analogy of this is. I'm a believer this can actually make a difference in health. So with that personal, I hope you'll excuse the personal story but I hope it illustrates the point this can make a difference in health. With that I'm going to end and we'll see whether we have some questions from here or the audience, Chris.

CHRIS DeGRAW: We do have several questions and a couple of comments that you may want to—

DR. ALAN GUTTMACHER: The comments would be.

CHRIS DeGRAW: Respond to.

CHRIS DeGRAW: First question is what effective public health strategies have been used to educate individuals as to effective use of the family health history after completing it? Also, have there been any negatives or down sides expressed by completing the tool?

DR. ALAN GUTTMACHER: Let me talk about the down sides first. Those are very important. I think the obvious down sides are a couple. One is the question of privacy and confidentiality. That's the question in a number of ways. First of all, any individual filling this out would want to know who is going to have availability of this. This was designed originally so that it never sat on a government server or anything else and it's clear it goes directly to the individual so only the individual filling it out has the availability to it. To make it that -- that led to technical problems with the downloadability. It will be changed in a way it will live in a government server for a millisecond. We have talked about this the same way the internal revenue service does things that are private, etc. There is a feeling that in fact the public will be OK with that. We'll have to put -- clearly there will have to be caveats there. The first thing is privacy. The other concern would be not so much if filling it out. The real privacy concern is once you put it in the medical record what happens to the family history? We think the biggest cure for that concern would be to have federal legislation that limited the use of genetic information in terms of its use by health insurers and by employers. There has been such legislation pending since 1996. It was passed in the first session of this Congress in the Senate unanimously, 98 to 0. The administration said they'd be eager to sign such legislation if it came to the president's desk. It is in the house currently. We have hopes that that would pass. That would probably help with that major concern. I think another concern, another downside

to this is, what is the psychological impact of knowing family history? And like any information about families, it can have psychological impact of many sorts. Looking at who has certain diseases. There is still this guilt of having certain diseases in our culture. There can be guilt within families about being the one who passes on the disease. Those kinds of things come into the open when you do that. I think that's one potential downside. We think there is an upside of getting it out and families talking about it. It has more an upside. What efforts have there been public health campaign to educate the public how to use this information once they have it? And that is something again that we have done a little bit about in this initiative but we hope to be doing a lot more about. To really primarily the major guidelines we're giving people today, once you have the family history, take it in your healthcare provider and talk with your healthcare provider about the family history, to begin that conversation, which does not happen enough currently in healthcare. Our real guide to how you use it, don't use it on your own. Gather the information but then take it to your healthcare provider to both refine the information but to most importantly have a good conversation in terms of the healthcare team, you know, partnering with the individual to figure out how to use that information.

CHRIS DeGRAW: Another viewer comments, we need to be sensitive to new or unconventional definitions of family where there may not be genetic or biological history. Not all people have access to their biological history.

DR. ALAN GUTTMACHER: A very important point and one you'll be glad to know that Michele Puryear made repeatedly as we had conversations about this last fall and many

of us were aware of this. And it is one of those things that in the newer versions of the tool we have a little bit up there about adoptive families but not enough on the website currently. We also will be giving guidance about how, if someone is adopted, what kinds of steps one might do to help gather adoptive history. It is important because it's not all about shared genes. They're biological phenomenon but also shared other kinds of things that have to do with step siblings, other kinds of non-biological relatedness. It's very important. As we get this more sophisticated and more complex in some ways there is some in there already but we'll be adding more attention to this because it's absolutely an important point. There are different kinds of validity but still validity there.

CHRIS DeGRAW: Is it only important to capture clinical diagnoses such as heart disease or also exposure to environmental factors such as second hand smoke, high fat diet or other factors that may led to clinical health problems?

DR. ALAN GUTTMACHER: It is still important. Less important but still important. It's the interactions of genes and environment. To some degree it's important that for instance if you had a family member that died of lung cancer, it would be very important to know, did that person smoke? If they smoked, it's less likely there is a hereditary element involved. If they didn't smoke it's more likely. If they didn't smoke and never worked as a bartender or waitress where they had a large environmental exposure or never worked in a smoke-laden environment and co--- we have to take the lung cancer more seriously. In another way looking at those things that we know cause disease so absolutely, if someone has a family history of lung cancer and they're a smoker then they have even more reason to be

concerned than if they have a history of lung cancer and not a smoker. The environmental factors come into play.

CHRIS DeGRAW: How will it be that family history become so complex.

DR. ALAN GUTTMACHER: There is discussion there about when to refer to a genetic counselor. It's homework we need to do. This is in the planning stages for this initiative if we can get the resources together to be able to do this, to create some computer-driven tool to help the family gather the information, we would like to have computer-driven tools that help health professionals analyze and utilize the information more effectively. But also, that part of that is to knowing when to refer. It's largely a professional education kind of thing. I think that's the job of the genetics professional community but not solely for us to talk about what are the reasons for referring. Now, many healthcare providers have in their mind reasons for referring for all kinds of things. When to refer to infectious disease specialist or anybody including when to refer to a Genetist. This is something we talk about genetic specialists being available. Letting people know how to find a genetic specialist in your area but it is important and as it gets more comprehensive we want to do it. I want to also say very clearly as genetics becomes pertinent to healthcare. When we look at the ten common causes of death, when we begin to understand the genetic factors and be able to stratify and individualize care, genetics becomes applicable to all our patients. We can't refer all our patients to genetic professionals. There are about 1,000 M.D. Genetists in the country. 2,000 genetic counselor. A few hundred nurses. It's not enough to take care of 300 million people. It will be -- it will have to be primary care

providers who do increasing levels of complexity. There still will be families so complex, individual patients with genetic testing results for families where they're so complex it will require referral to a genetics professional just as there are some problems that a pediatrician needs to refer. An ear infection you don't have to do that. The bread and butter genetic stuff we hope health professionals won't need to do that but somewhere they will clearly need to refer.

CHRIS DeGRAW: Discussion about adoptive families. The person comments an annual family history day would not be an easy or pleasant experience for adoptive families focusing on their lack of biological connection and increasing their awareness.

DR. ALAN GUTTMACHER: I don't think it need to be the case at all. I think those of us who have clinical or personal experience with adoptive families know that adopted members of the family are very eager, sometimes even more eager than the other folks in the family, to know about family. And are very eager to be part of that conversation. That's the thing. They need to be part of the conversation. As Michele said in the introduction this isn't just about shared biology. It's about shared environment and about culture and the family. If this conversation is done well so it's enjoyable for people it won't just be about -- it's impossible to talk about uncle George's lung cancer without talking about uncle George. In talking about uncle George, make it about a family event. An occasion to tell the families to bring the things out. If it's done thoughtfully and well the adopted members of the family can feel they're honored invites to this family feast and their family story is part of the family story now.

CHRIS DeGRAW: Have you considered developing a version of this tool to collect family history from the birth parents to be provided to the adoptive parents?

DR. ALAN GUTTMACHER: We've considered it but haven't implemented it yet. This gets a lot of attention. The whole adopted issue. It tends to get a lot of attention. Adoption is not rare in our society but it is not that common. It is interesting that I think we get a higher percentage of the questions we get about the initiative are about adoption than adoption as a proportion of individuals in our society. Clearly that is the kind -- we would like to have a menu of tools available here. We would like to have tools that are focused on talk about -- more on children's health than this one is. We want a menu specifically for adoptive situations so people can know what information would be most useful to get from the biological families. It's a very good idea, that kind of stuff.

CHRIS DeGRAW: Is there a list of medical disorders that we can suggest patients consider other than the six included in the tool as they complete the family health profile?

DR. ALAN GUTTMACHER: It's interesting. The six are interesting. They've been all vetted by having AHRQ and other related evidence-based medicine. Once you move beyond that six it's harder to draw the limits. And so that's -- we're trying to figure out exactly how to do that kind of thing. To give people points, these are other things you might want to talk about without saying there is other -- you don't want to talk -- the one thing you want to think about are single genes disorders, like cystic fibrosis, sickle cell.

Any one of those is relatively rare. Cystic fibrosis. The question of how we get the single Gene disorders, people pay attention to that is another one. We try to figure out how to do it. Eventually because it's web-based we can make it reacher by making it deeper and deeper. It will be a process. As we have more resource and time to build it those are the kinds of things to build into it those kinds of lists. Presented not just a list of things in an interactive way to lead people to these other things.

CHRIS DeGRAW: For our final question, what major would you advise for a college-bound student interested in pursuing further study in this field?

DR. ALAN GUTTMACHER: There are a couple possible. Science and biology would be an important major. Some undergraduates majoring in genetics these days. The wonderful thing about the use of genetics in medicine is I'm not sure I could imagine an undergraduate major that wouldn't be applicable to this. Right now we are working to develop a D.V.D. aimed at high school audience about careers in genetics and genomics. Biology makes sense, psychology. How do you explain it to people so they change health behaviors based upon it? Sociology, philosophy, the ethical questions about how this is applied and how people should do it. Pre-law because there are legal questions about should genes be patented. All these kinds of things. Engineering, bioinformatics for sure. Yes, go to college is what I would say and we'll be happy to walk you to the workforce when you get out.

CHRIS DeGRAW: I want to thank Dr. Guttmacher and Dr. Puryear for the interesting presentation. Thank the University of Illinois School of Public Health for making this technology work so well. Today's webcast, as with all of our mchcom.com webcasts will be archived and available within a couple of days on this website, "Come Near To Me," I know we have a lot of interest outside world in this webcast because we have a lot of people who wanted to participate today who weren't among our state directors group. So I think the archive is going to receive a lot of hits. We encourage you to let your colleagues know about the website and hope they'll find the webcast useful. We want to make these webcasts as responsive to your information needs as possible. If you have suggestions for topics you would like addressed on future webcasts or have comments in general, email them to us at info @ "Come Near To Me." We look forward to your participation again next month. Thank you.