COMMONWEALTH OF PENNSYLVANIA
HOUSE OF REPRESENTATIVES
PROFESSIONAL LICENSURE COMMITTEE

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PUBLIC HEARING
IN RE: HOUSE BILLS 2137 & 2138
LICENSURE OF GENETIC COUNSELORS

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BEFORE: MICHAEL STURLA, Majority Chairman
WILLIAM ADOLPH, Minority Chairman
Craig Dally, Julie Harhart, Susan Helm, Thomas Killion, Mark Mustio, Timothy Solobay, Richard Stevenson, Tom Yewcic and John Yudichak, Members

HEARING: Tuesday, July 23, 2008
Commencing at 2:00 p.m.

LOCATION: Radnor Township Municipal Building
301 Iven Avenue
Wayne, PA

WITNESSES: Reed Pyeritz, Kathy Valverde, Elizabeth Gettig, Rebecca Carr and Cristy Hollin

Reporter: Gina Brower

INDEX

PROCEEDINGS

CHAIRMAN ADOLPH:

Good morning. The hour of nine o'clock having arrived, I'd like to call to order a public hearing of the House Professional Licensure Committee. My name is Bill Adolph. I'm the Republican Chair, and today, I guess there's traffic and there's still some members of the Committee on their way. Representative Killian, who is the prime sponsor of the legislation that we're going to be discussing today, is probably caught in traffic. So without further ado, I'd like to start to have the members of the Committee identify themselves and the area of the state that they're from, starting with my far left.

Yesterday we all talked into these mics that were in here. As luck would have it, they were taken out to the shop last night. So we're going to be passing around these portable mics, okay, for everybody up here. The mic at the podium there is working. I want to remind everyone that this meeting is being filmed and it will be shown locally and possibly on PCN at a later date. Okay?

REPRESENTATIVE MUSTIO:

Thank you, Mr. Chairman. Mark Mustio,

from Allegheny County, the 44th District.

REPRESENTATIVE SOLOBAY:

Tim Solobay, the 48th District in Washington County.

REPRESENTATIVE YUDICHAK:

John Yudichak, the 119th District in Luzerne County.

REPRESENTATIVE HARHART:

Julie Harhart, the 183rd, Northampton, Lehigh Counties.

REPRESENTATIVE DALLY:

Craig Dally from the 138th District, Northampton County.

REPRESENTATIVE STEVENSON:

Good morning. Dick Stevenson, 8th District, Mercer County.

REPRESENTATIVE HELM:

Sue Helm, 104th District, Dauphin County.

CHAIRMAN ADOLPH:

And the nice-looking gentleman walking around with the mic is Wayne Crawford, the Republican Executive Direct of the Committee. Okay.

First item on the agenda --- the first testifier was supposed to be Representative Killion, and I'm sure he's on his way. When he gets here, he'll have some remarks. I'd like to bring to the mic the first testifier, Reed Pyeritz. How did I do, Reed?

DR. PYERITZ:

I accept just about anything.

CHAIRMAN ADOLPH:

Okay. Reed is the chief of the University of Pennsylvania School of Medicine, Division of Medical Genetics. Good morning.

DR. PYERITZ:

Good morning to all of you. Thank you very much for the opportunity to present my written comments, and I'll summarize these briefly. I live within walking distance of this building. My tax dollars built this beautiful facility. I will certainly recommend that they provide more parking out front for visitors. So my job at the University of Pennsylvania, and my previous lives in Pittsburgh and in Baltimore, is as a medical geneticist.

As a physician, trained in internal medicine and medical genetics, I provide services specifically to individuals who either have or have concerns about a genetic condition, be it a well-defined syndrome due to a chromosome abnormality or to a change, a mutation, in a specific gene or in any of the myriad common disorders that affect all of us eventually, that clearly have genetic predispositions, and which, in many cases, will run in families.

I would like to spend my professional time making diagnoses, recommending treatment and seeing patients in follow-up. But to provide complete medical genetic services requires considerably more than that. It involves close analysis of the family pedigree. It involves counseling individuals in many aspects of heredity and of how disorders, even within a single family, are quite variable and in unpredictable ways can affect individuals. Age dependency is a major aspect of this. I rely heavily on the genetic counselors with whom I work. I could not begin to perform my services without them.

Much of what they do, they do independent of me, with regard to specific clinical encounters with patients. Much of their work is done before the patient actually sets foot in the clinic, in terms of phone interviews in order to obtain detailed information about the family. Once I finish with the clinical business in the examining room, my counselor takes over and finishes the visit by dealing with the myriad of issues that remain, many of them psycho-social. Some of them quite practical, such as how to get a specific genetic test arranged and performed, and most importantly, paid for by their health insurance. They then deal with the results when they come back and notify the patient. All of this makes it possible for a seamless presentation of the complete clinical picture to the patient and the family.

Now the practicalities. There are really three kinds of providers of medical genetic services. There are physicians who are trained in clinical genetics. I give the exact numbers in the written testimony. About 1,200 of us have been certified by the American Board of Medical Genetics, since that came into existence in 1982. Many of us certified since then are not performing clinical services, for a variety of reasons that I numerate. So the actual cadre of clinical geneticists in this country is much smaller than the 1,200 or so who have been certified. Genetic counselors were certified from exactly the same point by exactly the same body, the American Board of Medical Genetics, for reasons too arcane to go into now.

The American Board of Genetic Counseling was created in the early 1990s and took over the certification of genetic counselors. So now there are two separate credentialing bodies.

There's also a group of professionals in addition to genetic counselors and clinical geneticists, genetic nurses. These are RNs who have training in genetics, and could well be considered to fall under the scope of the proposed legislation that we're considering today. There are many fewer of them than the 2,000, 2,500 or so genetic counselors who have been certified over the past 25 years.

The main limitation that we face in providing medical genetic services is that genetic counselors cannot be reimbursed for their services, unless there has been some specific contractual arrangement made with an insurance company. And these do exist, but are very few and far between. And the vast majority of the major payers, certainly in the state of Pennsylvania, do not accept such contractual arrangements.

So despite the fact that the average encounter, and we have published data that shows what the average encounter for my patients are, three hours, the only thing that gets billed and reimbursed is my time. And that is limited by the fee schedules. And it amounts to a hundred dollars, for three hours of work, much of which is provided by the genetic counselor. There are many instances when the genetic counselor can provide services independent of the physician, and should be able to be reimbursed for that.

Genetic counselors can bill. There are specific CPT codes in existence that identify genetic counseling as a service. But in fact, very few, if any insurance companies recognize genetic counselors as licensed, except in the states in which they are licensed, and hence, will not reimbursed for those billed services. Anything they do has to be billed under my name as the physician. So those are the logistics.

The practicalities are that you can’t pick up the paper most weeks without seeing something exciting coming out of the world of human genetics and genomics. The human genome project is seeing to that. We have a tremendous amount of new information to apply in the clinical setting.

The average physician trained at a time that genetics was in the backwater. And we know from many sources that the average practicing physician today really knows very little about genetics and how it can be applied in the clinic setting. They recognize this as well. They rarely overstep what they don’t understand, and hence, they rely on us to provide these services. And there are far too few medical geneticists like myself to deal with this. Increasingly there is direct-to-consumer advertising by commercial operations that are providing genetic testing. One opened up here in Philadelphia just a few months ago, called Smart Genetics, that will provide testing for a specific change in the AOE gene, that predicts what your risk will be for developing late onset Alzheimer's Disease.

This is something, a test that has been around for a while. But the professional societies have recommended against testing, first of all because it's not a sure thing, that if you have this genetic change you will get Alzheimer's Disease. And second, there is nothing we can do at this point and time to alter that risk in terms of medical therapy. Nonetheless, this is being offered on the web and in advertising to patients.

Any patient could send off a sample and have this information relayed to them. They then go to their physician and say, huh, look at this, tell me about this. And of course the physician has probably very little information with which to deal, so that then comes back to us.

This is the perfect situation where a

genetic counselor could do all of the work in terms of counseling the patient about results.

Direct-to-consumer advertising exists for the breast cancer susceptibility genes, for example. And those genes in fact do have tremendous clinical application at the present time. But it doesn’t take me to interpret those results and translate them into lay language for patients and their family members about what the risks are.

So these are just a few of the examples of where the need for genetic counseling services is growing exponentially in medical practice today. And the only way that this will be served in a reasonable way is if genetic counselors can be paid for what they do based on their specific services.

I’ll stop there and be glad to answer any questions.

CHAIRMAN ADOLPH:

Thank you, Doctor. We have a question from Representative Mustio. And I’d like to acknowledge the presence of Representative Tom Yewcic, who has joined us.

REPRESENTATIVE MUSTIO:

Thank you, Mr. Chairman. And thank you, Doctor, for your testimony. It kind of hits home a little bit. I, a couple of years ago, received a phone call from my sister, who has been diagnosed with breast cancer. And my mother had died of breast cancer, and her mother died of breast cancer. And she subsequently was treated and had a double mastectomy. And I received a phone call that said that you probably should consider having a gene test done. Which we ended up paying for to do.

And then the --- where the counseling I think comes into play is, do you want to do it? Do you want to know, first of all? And then once you do know, what do you do about it? So in my particular situation, tested and did not have the genes, so the likelihood of my children was a lot less. But unfortunately in her case she did and both of her daughters do, which I think increases their chance of breast cancer by about 80 percent, or somewhere in that neighborhood. So I think what we’re talking about here is that how do they then, as 21-to-24 year olds manage the rest of their lives? Marriage? Do they have kids earlier? Do they have kids at all?

Do they have --- I don’t know what the correct term is, but preventive mastectomies? And then where does all that get paid for? You know, people with individual resources can do that. Others that don't have the proper health care maybe cannot afford to have the test done in the first place. Is a genetic test $300? I don't know, is that roughly the number?

DR. PYERITZ:

Well, if the mutation is known in the family, you're looking only for that specific change. So yes, it's in that range. But defining what the mutation is the first time in the family, is $2,500 or so.

REPRESENTATIVE MUSTIO:

So it would be a lot more expensive. And we run into those issues of whether we want to mandate coverages and those type of things. And I don't know if I necessarily have a question, other than I wanted to verify that this is sort of what we're talking about here.

And then secondly, it seems like we tend to manage --- the terminology managed care came out originally where we're trying to prevent the expenses down the road. But now I think we're trying to manage premiums and not necessarily manage care.

And the way that we as legislators make the decision that this is a good investment to do up front. We just had a debate recently with covering colonoscopies. But I one, wanted to thank you for your testimony, and thank Representative Killion for the legislation. And just really share a personal story, but also from the standpoint that this test is very, very valuable to help people make decisions and also to give some peace of mind.

<u>DR. PYERITZ:</u>

Thank you for your comments. They've raised a number of important issues. And if I might just comment on two of them that you raise. The issue of managed care is an interesting one. In the staff model HMO, such as Kaiser on the west coast, they have actually hired genetic counselors to perform staff functions. So they are --- they recognize that within an HMO model, it's extraordinarily important, in a preventive sense, to have genetic counselors available for any of the array of services that are enumerated in this pending legislation. So unfortunately in the non-staff model HMOs, which are popular on the east coast, they don't reimburse for genetic counseling.

The second point that I'd like to emphasize that you raised is that genetic testing is unlike other medical testing. That when you have your blood count checked, basically you hold out your arm and you get a result in 12 hours or whatever it is.

And you sort of understand what your hematocrit is. Right? And your physician certainly does. But genetic testing carries many additional burdens, not only to the patient, but his or her relatives potentially, as well as to the physician who has to interpret the results.

And in fact, it's not a simple matter of holding out your arm to have the blood sample taken. There's a lot of pretest counseling that has to be performed so the individual understands what the implications are of the result. Be the result no mutation, definite mutation, or a change that we don't know what it means, in a gene. And there are plenty of those that come out as well, so that the need for post-test counseling is very intense.

REPRESENTATIVE MUSTIO:

Mr. Chairman, just one follow-up to that, the toughest part was deciding whether you wanted to know or not. Then once you've supplied the --- I assume it was blood --- then the anxiety of walking in to hear the results was really, really tough. So you provide a valuable service, thank you.

CHAIRMAN ADOLPH:

Okay, thank you. Are there any other questions for the doctor? At this time I'd like to acknowledge the presence of the Democratic Chair, Mike Sturla from Lancaster County. And I believe, Mr. Chairman, you have some questions.

CHAIRMAN STURLA:

Yes. I apologize for getting here late, but a couple of questions. One, in your testimony you talk about a genetic nurse and then a genetic counselor. Can you describe the difference between the two?

DR. PYERITZ:

Sure. A genetic nurse is someone who trained in nursing and has an RN certification. But who also has done additional training in genetics and provides genetic services. She or he may not have actually gone through a genetic counseling training program, and may or may not be certified as a genetic counselor in addition to being a nurse. This is a relatively small group, less than ten percent of the total pool of genetic counselors.

CHAIRMAN STURLA:

Okay. So you're saying less than ten percent of genetic counselors are also genetic nurses?

DR. PYERITZ:

Correct.

CHAIRMAN STURLA:

And you talked about some of the insurance implications. By licensing genetic counselors, do you think that would help in terms of getting the insurance reimbursement for this service?

DR. PYERITZ:

Absolutely. The insurance companies uniformly have said that --- they may come up with other reasons eventually as well not to reimburse, but certainly the first line of defense has been these aren't licensed health professionals. Hence they won't reimburse for their services. Even though they are certified by professional boards, they are not licensed.

CHAIRMAN STURLA:

Okay, thank you.

CHAIRMAN ADOLPH:

Thank you. Any other questions? Yes, Representative Stevenson.

REPRESENTATIVE STEVENSON:

Just to follow up on Chairman Sturla's question, are these --- are all of the genetic counselors, do they all have the same education, the same degree, a graduate degree and so forth? Is it similar or is it exactly the same for each?

DR. PYERITZ:

It’s very similar. Certainly for the ones who are being trained today, it’s very similar. Some of the subsequent people who will testify are actively involved in these educational programs, and can tell you how many there are in the United States. And generally they are two-year programs leading to a Master of Science degree in Genetic Counseling.

REPRESENTATIVE STEVENSON:

And then beyond the degree, is there additional continuing education in the field, or do they stop after that?

DR. PYERITZ:

No, no. There is a requirement for continuing education and for recertification or maintenance of certification, just as physicians.

REPRESENTATIVE STEVENSON:

In order to maintain their certifications?

DR. PYERITZ:

Correct.

REPRESENTATIVE STEVENSON:

Thank you very much, Doctor. Thank you, Mr. Chairman.

CHAIRMAN ADOLPH:

Okay, thank you very much for your

testimony and enjoy your walk back.

DR. PYERITZ:

Thank you.

CHAIRMAN ADOLPH:

At this time, I'm just going to do a couple of housekeeping things and then turn it over to Chairman Sturla. I was asked to submit into the record testimonies and letters submitted by the American Board of Genetic Counseling, American Board of Medical Genetics, National Society of Genetic Counselors and the Pennsylvania Department of State. Okay?

Without further ado, Chairman Sturla, the mic is yours.

CHAIRMAN STURLA:

Thank you. Next on our agenda is Kathy Valverde. She's a member of the PA Genetic Counselors Licensure Committee.

MS. VALVERDE:

Good morning, thank you very much for this privilege and the honor to speak today on behalf of the Genetic Counseling Licensure Committee. I represent the genetic counselors in the Commonwealth of Pennsylvania on a daily basis. I run a genetic counseling training program. There are two in the state. And I run the one in the Philadelphia area. I've been a genetic counselor almost 20 years now. And have sort of seen the evolution of the field. And I want to give you an understanding of what we do in genetic counseling. Genetic counselors are health care professionals. We all have specialized graduate degrees and training in medical genetics and counseling.

When we train, I sort of talk about having a three-prong approach. We learn about counseling, medical genetics and we do a research project. So all of us are trained in that way. We are part of a health care team. We provide medical genetics information and support to families who have genetic conditions and who are at risk to develop genetic conditions. Genetic counselors research the condition present in the family, interpret information about the disorder, analyze inheritance patterns, provide recurrence risk information and review available options for both the family and the care providers.

And as was mentioned today, genetic testing is different from individualized testing, because once you identify one person in the family who has a condition, there are implications for their other family members. So sometimes we see extended family members in our practice.

So why is genetic counseling licensure important? Well, it's a guarantee of minimal qualifications for our practice. Currently the public is generally not aware of the minimal standards for genetic counseling. This state regulation of the profession will ensure that genetic counselors are adequately trained and competent to provide genetic counseling services.

At the present time, anyone could basically call themselves a genetic counselor. Title protection limits individuals from calling themselves genetic counselors, unless they meet the standard and qualifications established by licensure.

And we want a guarantee of quality of service. Quality genetic counseling services are very important to the families who seek them. Genetic counselors in Pennsylvania are not required to take the national certification exam offered by the American Board of Genetic Counseling. Most genetic counselors work in hospitals. And the hospitals and the people that hire them usually ensure that they have that standard, but it's not required.

Individuals who have demonstrated

competency by qualifying and passing the national exam are less likely to commit errors that result in harm to the consumer of the service that is provided. Mistakes in practices will inevitably happen, but regulating providers through competency guidelines will reduce their frequency and help to avoid the emotional and economic costs of errors occurring in practice.

We also need to talk about accessibility of genetic services to the public. Genetic counselors have a strong and extensive knowledge in medical genetics. And their expertise in invaluable to members of the health care team.

Patients who receive inadequate or no genetic counseling service may have unnecessary testing or inappropriate testing, and may not be provided with access to the quality of care that they deserve.

And licensure would be the first step towards making genetic counseling more acceptable to the general public. This is especially important in the state of Pennsylvania, because there are a multitude of genetic counseling services available here in the Philadelphia area. The same is true in Pittsburgh. But if you go to the middle part of the state, and the more rural areas of the state, there are a relatively huge amount of counseling services provided. And these patients have to travel great distances to get the services they require and need.

Then there's legal recourse. Patients currently don't have legal recourse. If they are harmed by a genetic counselor practicing in Pennsylvania, without state regulation, incompetent genetic counselors cannot be punished by the state or restricted from practicing. And consumers cannot call a state agency to record a complaint against a genetic counselor or to check whether or not a person is an adequate provider of services. They could go to a web site and look at people who are certified under the American Board of Genetic Counseling, but that would take some knowledge of knowing how to get there. And currently we don't have a mechanism for most people to understand that.

So why is genetic counseling important now? Why is licensure for genetic counseling so important now? Genetic counseling is a relatively new field. There are only 29 genetic counselor training programs in the country. We've only been an established field about four years.

When I trained as a genetic counselor in the early 1980s, there were relatively few genetic testing options available. I very often only saw patients who had single gene disorders or very, very rare disorders.

And I talked to them about whether or not they would have a yes or no answer, yes or no are you going to have this genetic condition or not. And what Dr. Pyeritz was talking about was all of the advances today in technology, that now allow genetic testing for very common disorders, for common complex disorders. And these tests provide information about the level of risk, and sometimes they're very difficult to interpret.

So many of these tests that are now available directly to consumers, and consumers and genetic testing companies provide results, but they don't necessarily provide genetic counseling, nor give patients an understanding of where they can go to get appropriate counseling to understand the results of the testing that they had. And state licensure of genetic counseling provide consumers with access to highly-trained individuals to help with interpretation of counseling and test results.

I would just like to make sure you --- I provide this testimony in order to say that licensure for genetic counseling will help ensure a reasonable practice of our services in the state of Pennsylvania; hopefully protect the consumer; provide a mechanism to resourceful consumers if they are harmed; provide greater access to care for patients and families in the state.

And just as physicians and nurses have board certification and state licensure, we are seeking the same.

CHAIRMAN STURLA:

Thank you. Questions from members? Representative Adolph?

CHAIRMAN ADOLPH:

Thank you, Mr. Chairman. Thank you for your testimony. I'm not sure if Dr. Pyeritz had mentioned how many genetic counselors we have right now practicing in Pennsylvania.

MS. VALVERDE:

Well, I have a --- I've been on --- I've searched databases in states. Currently we have about 180 genetic counselors in the state of Pennsylvania. Which makes us the state with the third largest number of genetic counselors. California has the most, with a little over 200. Then New York is about 200, and we have 180.

CHAIRMAN ADOLPH:

Which leads me to my next question, Kathy. Are there any other states that are licensing genetic counselors?

MS. VALVERDE:

Uh-huh (yes). Currently the state of Utah was the first --- California was the first state to obtain licenses, to have a licensure bill. They have not issued a license actually for genetic counselors. The State of Utah was the second state. They have licensure for genetic counseling. And there are four other states that have licensure, Massachusetts, Illinois, Tennessee and New Jersey just passed a licensure bill.

CHAIRMAN ADOLPH:

Okay. All right, now the House bill that we have, House Bill 2137, and I just got word that Representative Killion is on his way. Okay? Is this a --- it this, the qualifications and the education background, is this similar to other states who have passed it?

MS. VALVERDE:

It’s very similar. We were fortunate to have states that went before us, so we could get an understanding of how they implemented their licensure bills. And so we looked very carefully at the Illinois licensure bills and the State of New Jersey. And they very similar.

CHAIRMAN ADOLPH:

Okay. Thank you.

CHAIRMAN STURLA:

Representative Yewcic?

REPRESENTATIVE YEWCIC:

Thank you, Mr. Chairman. Can you talk a little bit about the level of certainty that the occurrence will happen when you find the mutation and a little bit about the liability if you counsel somebody and a mistake has been made? You mentioned there's no liability?

MS. VALVERDE:

What I was talking about is if you go to a laboratory, if you see a family, and this how we always start when we see a patient. We take a family history, where a patient has a list of symptoms. And we try to figure out ---.

CHAIRMAN STURLA:

Excuse me, could you talk into the mic so that the ---?

MS. VALVERDE:

What potential disorder we're looking at in a family. That’s what I meant by a single gene disorder. And if we know what we’re looking for, we find --- if we do a laboratory test and we find a mutation in a gene that’s specific for that disorder then we have a very high level of understanding of how to proceed with the patients.

What Dr. Pyeritz was talking about a moment ago was about currently there are now testing where you looking at whole human genome sequences. Where you look at potential risks of developing something potentially later in life. And you don’t get a yes or no answer. It’s a potential for a risk. For those type of conditions, we have a lot less certainty.

So it’s much more confusing to explain to a patient when that might occur and how it might occur. So if somebody --- for example, I recently know of a case in Lehigh Valley where a person was seen that had a diagnosis of cancer. But the oncologist didn’t understand that sometimes cancer is isolated; sometimes it’s part of a genetic syndrome. And that family actually would end up having a genetic syndrome. And by the time they figured that out, two more people in the family were diagnosed with cancer, and that potentially could have been prevented. So sometimes it's very important to get a true understanding of what's going on. And that family really has no recourse that they didn't get appropriate genetic counseling. Does that answer your question?

REPRESENTATIVE YEWCIC:

What degree of liability is there then if a mistake is made. Is there any liability?

MS. VALVERDE:

As I said, genetic counselors work in a team. So there's liability to the institution that they work for, liability to the physician they work for, and liability to I guess the person who is practicing, you know, the genetic counselor. But it's harder to prove incompetence when you don't have state legislature, you don't have licensure to prove what the minimal level of competence should be.

REPRESENTATIVE YEWCIC:

One further question. What degree of certainty is it that if you counsel somebody that has some genetic disorder, is that disorder going to affect their future in reproduction or other ---?

MS. VALVERDE:

As Representative Mustio was just talking about, if you have the breast cancer gene, one of the breast cancer susceptibility genes, your lifetime risk of developing cancer is much greater than the general population. You go from a one in seven or one in eight general lifetime risk of an average as having cancer, to an 80 percent lifetime risk of getting cancer or getting breast cancer or ovarian cancer. So your risk is greatly increased. So that's very certain.

We can tell you that with a pretty good degree of certainty that your risk of cancer is greatly increased. So what do you want --- what options do you want to take? Do you want to increase your surveillance? Do you want to have prophylactic mastectomy? Do you want to have other family members tested so they can have increased surveillance?

So there's a high level of certainty that there's something we can offer those families, or there's information those families definitely need.

REPRESENTATIVE YEWCIC:

I guess that leads to one final question. If someone is counseled that they are at low risk, and they end up with that disease or whatever, does that create liability for counselors?

MS. VALVERDE:

Well, let's take the breast cancer case.

That would be a good example. Whenever you come in for BRCA testing, we tell you that we're looking for one --- that only accounts for about ten percent of all cases of cancer. So you could be --- you could have a negative, or not have the BRCA, one or two indications, and still get breast cancer as the general population would. So people would have to be made to understand that. We're testing for one type, one cause of breast cancer, not for all the causes that exist.

REPRESENTATIVE YEWCIC:

That one type of cancer, I have a less than ten percent chance of getting it, according to what you counseled me, then I get that cancer. Does that mean I turn around and sue you? Is there protection?

MS. VALVERDE:

I think that if you did an adequate job with the counseling, you would understand that there are multiple causes for cancer. And having the breast cancer gene is just one cause for getting breast cancer. So the person would have to understand that they're still at risk for cancer. It's just that their risk now is much lower. It's not a zero chance of getting breast cancer. It's just a much lower risk.

REPRESENTATIVE YEWCIC:

Thank you, Mr. Chairman.

CHAIRMAN STURLA:

Thank you. Representative Mustio?

REPRESENTATIVE MUSTIO:

Thank you, Mr. Chairman. As a follow-up to your comment about the case at Lehigh Valley, and sort of tying it into Dr. Pyeritz, if I'm pronouncing that correctly, he had indicated that I think it was United Health care has counselors, genetic counselors on staff, kind of look at these situations. And I would assume that that is there to help prevent any further situations. Would having genetic counselors on staff with the health insurance company, have given another set of eyes or helped that particular situation in the Lehigh Valley?

MS. VALVERDE:

Probably, because we're much more in tune to not looking just at the individual. We look at how that individual fits into their family and other types of cancers and problems in the family. So we get a greater understanding if that person --- other family members might be at increased risk. We're much more in tune to not just looking at the individual, but looking how that individual fits into their family, the whole family picture. That's our training and our job to see where that fits in, so we can identify other people that potentially might be at risk. Most practitioners see individual patients, and they're just concerned with the individual patient that they're seeing at that moment, and not thinking where that person is in the context of their family or risk to other family members.

CHAIRMAN STURLA:

Representative Harhart?

REPRESENTATIVE HARHART:

Thank you, Mr. Chairman. I understand that there are --- did you say 180 counselors?

MS. VALVERDE:

Roughly, yes.

REPRESENTATIVE HARHART:

In Pennsylvania.

MS. VALVERDE:

Uh-huh (yes).

REPRESENTATIVE HARHART:

Okay. Out of those 180 counselors, are they all specialized counselors? Because you said in your testimony, at the present time anyone can call themselves a genetic counselor. Do they all have the training and the education to be a genetic counselor?

MS. VALVERDE:

The 180 that I have on my database list, that I've obtained that list by contacting all the hospitals in the state, looking through the National Society of Genetic Counseling registry, looking through the American Board of Genetic Counseling's list of certified practitioners, most of the genetic counselors on our list work in major medical centers in either prenatal genetic services, cancer oncology services, pediatric services, medical genetic services.

The vast majority, almost all of them, have trained through a genetic counseling training program. There are a few exceptions of people who trained very early on, who then went back and got certification as a grandfathered-in kind of a practice, and they were able to be grandfathered. But the vast majority have trained in the typical way, going to one of the 29 genetic counseling training programs in the United States, and obtaining certification with the American Board of Genetic Counseling.

REPRESENTATIVE HARHART:

How easy would it be to call yourself a genetic counselor? Because it sounds like you really do have to be very specialized. And if you're going to be hired in a hospital or any setting that has been brought forth, how easy is it to say you're a genetic counselor and not really be what you say you are?

MS. VALVERDE:

I guess my concern, it's not very easy if you worked in an institution where there were regulations regarding that. What's concerning me now is all this direct-to-consumer marketing and testing. So it's moving out. Genetics is moving out of our institutions. It's moving out of medical genetic services provided by major medical. You can go and you can find services. I don't think it would be --- I know of cases of people who put web sites up and have said they provide genetic counseling services. That is a possibility, and it does exist.

You could go into like somewhat of a private practice working for a physician in a private office, and I guess you could call yourself a genetic counselor. It would be unlikely in a major medical institution like the University of Pennsylvania Hospital System or one of the majors, because they would understand the credentials and the qualifications.

The concern for me is that as a consumer, you wouldn't know what those qualifications and credentials are. So it would be hard for you to know whether or not you're seeing someone who is qualified or not. You can't check the credentials very easily.

REPRESENTATIVE HARHART:

Thank you.

CHAIRMAN STURLA:

Thanks. Representative Helm?

REPRESENTATIVE HELM:

Thank you, Chairman Sturla. You've mentioned that you've been doing this for 20 years, so I imagine you have an extensive background and history. And I was just wondering, the adopted child, like do you find adoptive people come to you with questions and how genetic testing would help them know, since they obviously know nothing about their biological family? I just wonder if you could talk to me a little bit about that.

MS. VALVERDE:

For the past few years, I haven't practiced as a genetic counselor in terms of seeing patients on a daily basis, because I run a genetic counseling training program. But when I did practice as a genetic counselor, that very often comes up regarding when people are concerned about having children. I used to very often see families who would come for pre-conceptual counseling and ask that question. You know, I don't know anything about my background, so I'm concerned about what potentially I could pass on to my child. I know there's certain carrier screening that we do based on ethnicity testing.

And people would come to see me and say you know, I'm Caucasian, but I don't know if I'm of Jewish ancestry or Italian ancestry. There's certain conditions that we look for through carrier screening based on those instances. So some of those families get a lot of testing, because they're anxious and concerned. So there is testing available, and we can help them interpret it. For the most part though, if a person is healthy, and we don't see any symptoms in them, the chance of passing something on --- but there are like late onset disorders that they wouldn't know about. So some states have protection where you can go back and look at your birth records. And some people have birth records that they can access, their family history records.

REPRESENTATIVE HELM:

Thank you.

MS. VALVERDE:

Uh-huh (yes).

CHAIRMAN STURLA:

Thank you. One more question.

MS. VALVERDE:

I'm sorry.

CHAIRMAN STURLA:

The concern that is often raised by members of this committee is so what harm gets done to the public if we don't license you? Can you explain what happens with somebody who gives bad counseling?

MS. VALVERDE:

I think there are two potential problems. The first potential problem would be the person doesn't get the testing they need. So if you come in with a family history of a certain condition, or you know --- say the example Representative Mustio gave, you have a grandmother who died of breast cancer, a mother who died of breast cancer. A person in that situation, a physician should say that's a strong family history of cancer. You should go see genetics providers who might discuss with you the potential for genetic testing and whether or not you're interested in having that.

Sometimes people get to that testing early enough to prevent the cancer from occurring.

Sometimes they get there too late and the cancer has already occurred. And we all know it's better to treat a cancer in early stages, rather than when it's progressed. The alternative of that is someone goes to a direct-to-consumer testing company and gets a test done. And they don't understand risk assessment. And they tell them, you know, the potential of having this problem and making life decisions and changes based on the potential of getting Alzheimer's when they're in their 80s, when that might not be true.

So you're making life decisions based on something that's happening why in the future that might not be very accurate. So there's importance of having testing to do for prevention and there's importance of having testing for reproductive choices, life decision making. So if people get erroneous information, they make bad choices. And we're trying to prevent that by giving people the most accurate information to make the best choices they can.

CHAIRMAN STURLA:

Okay, thank you. Next on the agenda is Elizabeth Gettig, Co-Director of the University of Pittsburgh's Genetic Counseling Program.

MS. GETTIG:

Good morning. Thank you for your time

and your travel here today. I am from Allegheny County. I am the Co-Director of the University of Pittsburgh Genetic Counselor Training Program. And like Kathy, I've been in the profession for quite some time, having finished my degree work in 1981. I'd just like to tell you a little bit about the training situation here in Pennsylvania. Two of the 30 accredited genetic counselor master's degree training programs are located here in the Commonwealth. Only two other states have two genetic counseling programs. And those are Maryland and California.

Most states don't even have a single counselor training program. Arcadia is the program that Kathy directs. And I direct the University of Pittsburgh program in the Graduate School of Public Health. Many graduates choose to stay in the Commonwealth. We have a lot of students who are from our general areas. And I think that we have saturated the Philadelphia, Pittsburgh, Harrisburg, Hershey, Danville area markets with Pennsylvania graduates quite a bit. And they are very happy being close to family and relatives. That's one of the reasons we have such a large number of genetic counselors in our state. But we also have students who go far away, but they would like to have knowledge of this concept of licensure in their state of training.

Licensure can create clear educational, certification, and continuing education standards for all genetic counselors. It will ensure greater patient access to competent genetic counselors. The goal is for patients to make more informed decisions about their genetic health, enabling them to seek methods to prevent disease and to stay healthy. Genetics is a complex science. It’s very daunting to most members of the public, that when you say genetics often they get an image of some sci-fi movie or some other event. And so we have to be very careful in how we approach families, because this is an area that is very emotionally sensitive to individuals.

And it also has the ability to affect multiple family members. So it’s not just you coming in for a consultation. It’s really like your whole family coming in with you as well. It’s important that we do make efforts to protect the public in this process. This legislation’s intent is to ensure that individuals and families who are being counseled on genetic information are provided that information by qualified and trained practitioners. This assures protection of the public.

As a genetic counseling program director and provider of services myself, I know that providing services to help families and individuals translate this very complicated scientific knowledge into practical information is a challenging task. A genetic counselor works with the person, or the family that may be at risk for an inherited condition. One of the lessons that we have learned from the human genome project is that we are all at risk for something. So it's not that any of us is immune to any type of genetic change. Some of these are relatively benign changes, but some of them can have medical consequences as well.

As tests become available for the public, it's our responsibility to assure accurate information is being provided by appropriately trained professionals. In order to respect your time this morning, I sort of presented my testimony in a question and answer form. So one of the questions that comes up is, do genetic counselors have licensure now? And as was previously addressed, no, in most states they do not. But I know all of you are familiar with the National Council of State Legislatures. And the NCSL web site does have the legislation from the other states that do have this type of legislation in place.

I’m formerly an elected official in Allegheny County. I was president of my borough council. And I must say I do have room envy very much, because our council chamber certainly was not even a mere reflection of where we are today. But with elected office, I know comes great responsibility.

Genetic counselors really do desire licensure. And in the states of California, Utah, Illinois, Oklahoma, Texas, now I hear New Jersey, Massachusetts, all have licensure regulations in place. So those are states that have even fewer genetic counselors than the Commonwealth does. And I think that these protections are very important for the public, so that the patients do have recourse if they get misinformation.

One of the case examples from the last testimony that I’d like to add is that I had a military family that I worked with, who had a baby with Down syndrome. Their baby had a chromosome picture of 47 chromosomes, with the extra one being the number 21 causing the Down syndrome condition. When you have trisomy 21, it means that you have a one percent chance for having a future child with this condition. In the military, this family was counseled that it wasn't a one percent recurrence risk, it was a 100 percent recurrence risk. And as a result, had no more children.

When I met them later, here in Pennsylvania, I said you know is that really how you recall the situation? Did they tell you that it was 100 percent? And that is the recall of the family. Now, what happened at that time of stress, when they got the diagnosis, and how people heard the information, I can't attest to. But I know that the husband had a vasectomy and the mom had a tubal ligation, because they were afraid of having another child with special needs. When we explained to them that their recurrence risk actually was one percent, they had reversal of both of those procedures, and went on to have three healthy children.

They didn't have any recourse to say, someone gave me poor genetic counseling, or someone gave me misinformation. And that's what this type of legislation would help us do. It would protect the public from misinformation related to genetic testing. Another question that came up earlier is that will this affect nursing scope-of-practice issues. The genetic nurses are already covered by their licensure. As you know, all RNs are licensed here in the

Commonwealth. So their scope of practice will not be affected by this. This would be legislation that is intended more for those trained in the master's level of genetic counseling process, those 180 individuals that Kathy referred to.

And how many genetic counselors are there here? Again, there are about 180. But we also have another large group, and those are those in training. So we have the second and third largest training programs in the United States here in the Commonwealth. So we do have a number of students as well. And they want to know, will I be licensed. Is it something beyond certification for us.

So that's the conclusion of my remarks. I'd be happy to take any questions.

CHAIRMAN STURLA:

Thank you. Questions from members? Representative Dally.

REPRESENTATIVE DALLY:

Thank you, Mr. Chairman. Thank you, ma'am, for your testimony. As part of your counseling, do you ever provide counseling to individuals that they should undergo genetic testing, or are you always involved after the testing is done?

MS. GETTIG:

What we do is help people through the decision-making process. As the representative indicated, we start with the first question, do you wish to have this information or not. We would not be what we'd call direct, and say take the test. Go get the test. You must have the test. We would say, the test is available. And this is what the test can tell you. Here are the benefits and here are the risks. And then the family or individual makes the decision whether or not they wish to pursue the testing.

REPRESENTATIVE DALLY:

And then after the results are received, then you provide the counseling?

MS. GETTIG:

Then we interpret the testing results to each individual.

REPRESENTATIVE DALLY:

Thank you.

CHAIRMAN STURLA:

Questions from other members?

MS. GETTIG:

And I'll just state for the record, that even though I have a southern accent, I am a native Pennsylvanian.

CHAIRMAN STURLA:

So how do you acquire a southern accent?

MS. GETTIG:

I grew up in North Carolina.

CHAIRMAN STURLA:

All right, Representative Yewcic.

REPRESENTATIVE YEWCIC:

Real quick. This is a licensing bill. Should there be any limitations for any type of abuse? I mean you've mentioned sci-fi and all these things. I mean I can imagine someone forming some kind of dating web site, saying go get your perfect match. Should there be limitations on ---?

MS. GETTIG:

I think it would be a limitation to rendering of health care related items.

REPRESENTATIVE YEWCIC:

Just for health care, looking for disease and genetic type ---.

MS. GETTIG:

Exactly. I don't think you should have genetic counseling for internet dating.

REPRESENTATIVE YEWCIC:

Well, I mean that will be out there.

MS. GETTIG:

Exactly. It will be. But I think really

what we want to confine ourselves to is health-related issues.

REPRESENTATIVE YEWCIC:

Okay, we don’t want someone out there looking for the super race and all that kind of nonsense.

MS. GETTIG:

No.

REPRESENTATIVE YEWCIC:

Thank you, Mr. Chairman.

CHAIRMAN STURLA:

Thank you. Questions from other members? Representative Adolph?

CHAIRMAN ADOLPH:

Just to get this on the record. Most of your patients, are they referrals from physicians?

MS. GETTIG:

From physicians and self referral.

CHAIRMAN ADOLPH:

Okay. Thank you.

MS. GETTIG:

Thank you.

CHAIRMAN STURLA:

Okay, thank you. Do you want to do your part now or do you want to wait ‘til the other ---?

REPRESENTATIVE KILLION:

I'll just say a quick word.

CHAIRMAN STURLA:

Okay. We'll go back to Representative Killion here for some comments.

REPRESENTATIVE KILLION:

Thank you, Mr. Chairman. Let me first start by apologizing. There's nothing worse than at 20 after 9:00 to look and see you have a voicemail on my way with my cup of coffee, to what I think I've got to be here at 10:00 is the Minority Chairman Adolph saying it's two minutes 'til nine, Tom, we're about to do your bills. So let me apologize for being late. And let me also thank Chairman Sturla for holding the hearings on House Bill 2137 and 2138.

I'm not going to talk too much. I'd rather just hear the rest of the testimony, and again, let me apologize for being late. But I do think these bills are important. I think it's important for our doctors and our hospitals to be able to recognize qualified genetic counselors. And licensure will do that. They do provide direct patient care, and I think it's important we protect the patients in Pennsylvania. Thank you.

CHAIRMAN STURLA:

Thanks. Next on the agenda is Rebecca Carr, the outreach coordinator for the Philadelphia Network for FORCE, which is Facing Our Risk of Cancer Empowered.

MS. CARR:

Good morning. My name is Rebecca Carr, and I am resident of Lafayetteville, Pennsylvania. I am a mother, a wife, a social worker and a volunteer. I am also a four year survivor of heredity breast cancer.

In the fall of 2003, I found a lump in my breast which turned out to be cancer. This was a shock to me, since no one in my family has had cancer in their 30s, like me. In between chemotherapy appointments, my medical oncologists had the good sense to send me to the genetic counselor at my hospital. The counselor mapped out my family tree with me and explained where she saw a red flag.

I was surprised to find that that flag was on my father's side of the family. We had to travel up to my great grandmother who died in her early 30s of what they called female cancer. What we probably today call ovarian cancer. This was enough to warrant genetic testing, which I did. About a month later, the results came back positive for genetic mutation, called BRCA-1. This mutation was the reason that I had gotten cancer. Harboring this mutation gives a woman around an 85 percent chance of developing breast cancer, and a 60 percent chance of developing ovarian cancer.

This information was at once devastating and liberating. On the one hand, I realized suddenly that everyone in my father’s genetic line was at risk. And on the other hand, I realized that by my getting cancer, and by my visiting with A genetic counselor and getting tested, those relatives would have the option of counseling and testing before they ever developed this terrible disease.

Others in my family have met with genetic counselors and been tested. Some have tested negative, and now know that they have no greater risks for cancers than the general population.

Others have tested positive, and are in a position to make pro-active decisions and have heightened surveillance so that they too, we hope, never develop cancer. In the meantime, I fought my battle, and along the way consulted two other genetic counselors at our finest Philadelphia area hospitals, as I struggled to figure out what my genetic mutation meant for my health, for the rest of my life.

These counselors were able to arm me with the information that I sought, so that I could make the best pro-active decisions for myself and for my family. I was fortunate to find the good people of FORCE, Facing Our Risk of Cancer Empowered, an organization which serves to improve the lives of individuals and families affected by heredity breast and ovarian cancer.

I am currently the co-chair of their Philadelphia outreach group. And I am proud to read a letter of support from their executive director.

On behalf of Facing Our Risk of Cancer Empowered, FORCE, and the community we serve, I support efforts to establish licensure of trained genetic counselors in Pennsylvania state. Our members consist of individuals and families at high risk for heredity cancer. We want to ensure that all interested individuals and their families have access to quality non-directed genetic counseling with qualified experts about their personal health circumstances and their decision-making regarding genetic testing.

Further, we endorse the establishment of competency standards to help assure that all health care providers offering genetic services have knowledge about the tests they are providing and the ability to order and interpret genetic testing appropriately.

The lay public has limited ability to determine who is qualified or adequately trained to provide competent genetic counseling. FORCE has compiled case studies of people who received incorrect or inappropriate information regarding their risk for cancer and genetic testing from health providers who are not properly trained in cancer genetics, and the unfortunate outcome when people make medical decisions based on incorrect or incomplete information.

Genetic counselors offer vital expertise for those seeking to learn more about heredity cancer and health. Through rigorous advance training, these professionals are uniquely equipped to provide expert guidance to patients and families.

Genetic counselors are also growing in number. As the use of genetic testing and other genetic services increases, the demand for genetic counseling is rising. The establishment of standards and licensure for this profession will help to ensure quality for patients and their families.

In establishing licensure standards, we support the requirements set forth in certification by the American Board of Genetic Counselors or the American Board of Medical Genetics as a minimum standard of practice for genetic counselors in every community.

We urge the passage of a licensure bill that will ensure that qualified genetic counseling can be obtained by all the citizens of Pennsylvania. Sincerely Yours, Sue Friedman, Executive Director for Facing Our Risk of Cancer Empowered.

So I am the patient. I am the one who meets with these counselors. I'm very open about my personal situation, and I'm happy to take questions.

<u>CHAIRMAN STURLA:</u>

Thank you. Questions from members?

<u>MS. CARR:</u>

No? Okay. Thank you.

<u>CHAIRMAN STURLA:</u>

Thank you. Oh, Representative Adolph?

<u>CHAIRMAN ADOLPH:</u>

I don't want to let her walk away. You may have mentioned this, your children.

<u>MS. CARR:</u>

Yes.

<u>CHAIRMAN ADOLPH:</u>

Now, have you thought about when you're

going to start testing your children?

MS. CARR:

It is the universal, I guess, advise from the genetic counselors I've spoken with not to test children until they're 18 at the very earliest. And even then, perhaps not, because it is a huge burden to know this. I mean think about your typical 18-year-old going off to college and starting on their own. And if they were to know this, that would be very hard. There's also very little they can do at age 18 to minimize their risk.

I have a boy and a girl. They each have a 50/50 chance of carrying this mutation. If they do not have it, it stops with them. They cannot pass it on. They have no greater risk. So 50/50 is what we have. And I sort of look at my children as if I'm going to assume they have it. I'll do everything I can so they have a healthy upbringing. They eat well. They exercise. They do what they can. And then we'll test them eventually. I hope when they are 18, 20, 25 maybe we'll have a cure for cancer and it will be a non-issue. That's my goal.

I want to take two more minutes, because my situation --- I'm a survivor. I'm a person who found I have cancer and then went to the genetic counselor.

My co-chair at FORCE is what we call a previver. And that's a hot new word. It's actually one of Time magazine's top ten words of the year last year. A previver is someone who has not had breast or ovarian cancer, but knows they have the genetic mutation for it. So they can take action. And my co-chair in FORCE is a previver. She watched her mother and grandmother die. She lived her whole young adult life knowing there was this hanging over head. And she went to Fox Chase Cancer Center, and went to their genetic counseling department and said, okay, I've had my children now. I'm ready to talk about this. What do I do.

And they were actually able to find her mother's tumor, frozen, from ten years earlier, and get that out and test it, because like one of the people said earlier, you don't want to be looking for everything that's out there. You want to zoom in on what mutation my friend probably has. So by testing the mother's frozen tumor, yes, they found the mother had BRCA-1. And they tested my friend, the daughter, and found that she too had BRCA-1. And she's had her children and she said I will not live with this cloud over my head. After genetic counseling and extensive soul searching, she had preventive mastectomies.

Called me the next day from the hospital, said Rebecca, I am so incredibly happy. I feel like this cloud is finally lifted from over my head. I'm going to be here to raise my children. And I'm going to live a long and healthy life. So those are sort of two ends of the spectrum in the breast cancer community of who uses the services of genetic counselors.

CHAIRMAN ADOLPH:

Rebecca, yeah, I'm sure everyone here probably has personal situations regarding breast cancer in their lives. Do most hospitals --- and I'm not sure if you should be the one answering this question, and maybe someone else can get back up. Do most hospitals employee genetic counselors?

MS. CARR:

Genetic counselors. The three hospitals that I have worked with in my cancer battle all had genetic counselors that were available to me, yes.

CHAIRMAN ADOLPH:

And that's --- and either a physician or a nurse in the hospital, are they the ones that referred you to genetic counseling?

MS. CARR:

Yes, my doctor, because I was young, because I was in my 30s, there was this was this sort of question, because that's not generally when we see breast cancer. So there was this question of why. And that's why I was sent to a genetic counselor who did this massive family tree and found this great grandmother. And that was the red flag. And that was enough to say okay, we want to test you, because we think there is something traveling through the men in your family. That by the way seems --- I'm not a professional, but of the mistakes that I've heard that are out there, and people who have gotten wrong information, the one that pops over and over again is, oh, it can't travel through the men. But it does. The men carry this mutation, too.

CHAIRMAN ADOLPH:

Okay, thank you.

MS. CARR:

You're welcome.

CHAIRMAN STURLA:

Thank you. Finally on the agenda is Cristy Hollin, parent of a teenage utilizing genetic counseling.

MS. HOLLIN:

Good morning. I am freezing here, I

don’t know if any of you are. But thank you for having me here today. My name is Cristy Hollin, and a long time ago I was a lawyer, in a land far away. But now I’m coming to you as a mother. And I’m the mother of a 15-year-old son with special needs and his name is Matthew.

When I was pregnant with Matthew, I interviewed three obstetricians and explained my unique family history. And I have two first cousins who are mentally retarded. Both males, and they have a typical female sister in between the males. Each obstetrician told me I have nothing to worry about and don’t require any genetic testing.

If, however, they had me consult a geneticist, my life would have been quite different. I could have explained to the geneticist the specific symptoms exuded (sic) by my cousins, including mental retardation, hand flapping, perseveration of speech, elongated faces, and guaranteed any geneticist worth her weight would have suggested that I be tested for Fragile X Syndrome on the spot.

It would have been a simple blood test, easy, efficient, cost effective. And then life for me and my family could have been quite different. I could have been armed with adequate knowledge to make choices and understand the future of me, and my family and my son.

But instead the obstetrician acted on their own limited scope of knowledge, and diagnosed me as having no risk of passing a genetic condition on to my own child. After Matthew was born, his symptoms as a newborn were slight, but present. It’s a painful time during those first early years thinking that something may be wrong with your child, but praying that you’re wrong. Early symptoms for Matthew included lack of eye contact, not reaching for toys, inability to sit up by six months and after, or even reach up to give me hugs. They mount to circumstantial evidence as something not quite right with your child.

Something you clearly want to ignore, but that continually nagged at you. During these early years we went to a number of specialists, none of whom suggested genetic testing. We went to DuPont Hospital, where we were told Matthew is probably mentally retarded. But then were shown the front door.

Picture two hysterical parents driving their little boy home from Delaware, their entire world shattered, but having absolutely no answers nor anyone to turn to. Next, we went to the head neurologist at a very prominent children's hospital in Philadelphia, who told us that Matthew was perfect, to take him home and love him.

And the list can go on and on of our adventures. And finally, our diagnostic journey ended with a simple phone call. My father-in-law had a phone conversation with a geneticist at the Elmwood Institute named Brenda Fenuken (phonetic). My father-in-law described Matthew's symptoms to Brenda. And in about one second had a possible diagnosis for Matthew. In fact, he probably had something called Fragile X Syndrome.

We immediately followed this up with a visit to the genetic department at Children's Hospital of Pennsylvania, where we received Matthew's official genetic diagnosis of Fragile X Syndrome. That's all it took, was a left turn down the right hallway and viola, we had our diagnosis. That easy.

The importance of the diagnosis cannot be stressed enough. It allowed me to do in utero testing, resulting in two unaffected brothers for Matthew. And allowed me to join a support group, learn specific ways to education Matthew, get him proper county and school district services, educate others. And my family alone has raised thousands and thousands and thousands of dollars to find a treatment or cure for Fragile X. And in fact, Matthew and I go to the Osteopathic Hospital on City Line Avenue every year and lecture the first year genetic students on the importance of genetic testing.

Of further note, the genetics department of the Elmwood Institute has spent countless hours with me and my family to help us understand Fragile X and the impact it has on our immediate and extended family. They've helped me understand genetics enough to slightly educate others. Fragile X has a complex inheritance pattern. And only with the help provided by an experienced geneticist, could I begin to educate myself and others. I really feel like geneticists are the Sherlock Holmes of medicine. They're giving clues and must use the specific information they've been taught to turn these clues into a proper diagnosis.

Their impact on their patients' lives is tremendous. For my family, it ended a tortuous search for Matthew's diagnosis, and allowed us to move on with family planning and educational success for Matthew.

To the obstetricians and other health care providers I encountered during these 15 years, I always wanted to tell them how inadequate they were. And that all they had to do was tell me to make a left turn, down the right hallway and into the genetics department.

That’s it. Thank you.

CHAIRMAN STURLA:

Thank you. Questions from members? Representative Adolph?

CHAIRMAN ADOLPH:

Cristy, thank you for your testimony, and to all of you who have testified. Do you mind taking some personal questions?

MS. HOLLIN:

We’ll see what you got for me.

CHAIRMAN ADOLPH:

Okay, fine. Is Matthew your first child?

MS. HOLLIN:

Matt is our oldest, yes.

CHAIRMAN ADOLPH:

Okay. And once you were able to find out what Matthew had, this Fragile X Syndrome ---

MS. HOLLIN:

Uh-huh (yes).

CHAIRMAN ADOLPH:

--- do you have other children?

MS. HOLLIN:

Two other boys.

CHAIRMAN ADOLPH:

What exactly --- once you found out what it was, when exactly were you able to do, to go ahead and have other children?

MS. HOLLIN:

Well, the first thing I did --- first of all Fragile X carriers have premature --- I found out all this other information, once you get in the loop, that Fragile X carriers have premature ovarian failure. So immediately my husband and I were very concerned about our reproductive options, concerning that, and the fact that I carry this genetic anomaly.

So I ended up traveling to Fairfax, Virginia and was part of this test group that was doing pre-implantation diagnosis on Fragile X carriers. So that was one thing that was not successful with me. But we ended up having an opportunity to become pregnant naturally. And the first thing that we did was get a CVS, which is your earliest way of determining whether or not the child that you're carrying has Fragile X.

CHAIRMAN ADOLPH:

Thank you.

MS. HOLLIN:

And with that, you could just make choices about what you want to do, yeah.

CHAIRMAN ADOLPH:

Thank you.

MS. HOLLIN:

Sure, thanks.

CHAIRMAN STURLA:

Thanks, any other questions? Okay, thank you. Obviously an issue that bears review of this committee, and we will look at moving this bill in the near future.

With that, I guess we will conclude the meeting. Representative Adolph?

CHAIRMAN ADOLPH:

First of all, Chairman Sturla, I want to thank you for holding this hearing here in my legislative district. I mentioned at yesterday's hearing, I've been in the legislature 20 years. This is the first time I've had a road show come to Delaware County, in my legislative district.

I want to thank the Commissioners of Radnor Township for providing this beautiful facility to us. This building was dedicated this year, and I wanted to show off a little bit. Not all the municipal buildings in Delaware County look like this. Okay? And the taxpayers of Radnor Township, thank you, okay, for providing revenue for building such a building.

For those that are taking off, I'm going to plug our local restaurants before you leave and get on the turnpike. Okay? Just make a left on Lancaster Avenue out there and you're going to see a good 50 to a hundred restaurants. And you have to try some of our specialties, the hoagies and the cheese steaks in this area are fabulous. Okay?

I want to thank all my colleagues for coming, and of course the testifiers today for sharing. I thought it was a very educational and informative hearing. Thank you very much.

CHAIRMAN STURLA:

Thanks. I also just want to point out, Representative Gergely is on his way. I guess he was going to the same ten o'clock meeting I was. That's what happens when we change the times on these hearings. And he's in traffic, but will be here. So with that, we will conclude the meeting. Thank you.

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HEARING CONCLUDED AT 10:16 A.M.

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