

## **AMCHP 2008 ANNUAL CONFERENCE**

### **WE ARE MAKING A DIFFERENCE:**

#### **Using Mapping Software to Identify Barriers to Special Medical Services**

March 1st to 5th, 2008

KATHARINE HARRIS: Hawaii has just been wonderful in terms of genetics and newborn screening stuff. Well, we'll just start. Okay? And I'm just going to do a demonstration using a mapping software that we did. But a little bit of introduction to why we did it, okay? And just obviously stop me anytime you want to. The objectives of this talk will be to identify the regional genetic and newborn screening service programs and their goals, identify the state newborn screening programs and the core conditions, understand some of the barriers to accessing necessary medical services, understand how mapping can highlight geographic disparities and just learn one software mapping program. Obviously there's lots of them.

HRSA, Health Resources and Services Administration--you're probably familiar with them, that's why we're all here--has funded the seven Regional Genetic and Newborn Screening Service programs and the goal, the vision or the goals of these regions is to improve the quality of health of children and help their families by promoting the translation of genetic medicine into public health and health care services and never quite sure, you know, all these words put together and

you're going, what does that mean? But take a regional approach to address the mal-distribution of genetic resources and the problems families and primary health care providers have in accessing and utilizing those resources. And that's really what we're hoping for.

The primary goal is sort of confused and in dispute, but as stated in the guidance is to ensure that individuals with heritable disorders and their families have access to quality care, appropriate genetic expertise and information in the context of a medical home that provides all the stuff that medical homes are supposed to provide. And you probably couldn't recite it as well as I can.

Objective is to strengthen communication and collaboration among public health individuals, families, primary care providers, genetic medicine and other subspecialty providers and to quantitatively and qualitatively evaluate outcomes or projects undertaken to accomplish their goals. Those are the objectives of these seven regions. And these are the seven regions—you guys are out in the western states. We're here in—this is NYMAC, New York Mid-Atlantic Consortium, I thought it should be Mid-Atlantic New York Consortium but when that talk says MANIC instead of NYMAC, we had a little confusion and disagreement. Let's see, I think NYMAC is this collection of states in New York, Pennsylvania, New Jersey, Delaware, Maryland, D.C., Virginia and West Virginia. I don't think I'm going into it, but I've been with these kind of regions since 1988.

And in 1984, actually, you might know of the earlier permutations of these regions (inaudible). I don't know if you were around. At that point, Hawaii was part of the south, Pacific South region with California and Nevada and nobody knew about Guam. So, it didn't play with them, but those were funded to 2000 to 2002. NYMAC was actually two different regions. NYMAC was the margin, the Mid-Atlantic Human Genetic Resources region and also the Genetic Network of the Empire State until we realized we also had Puerto and Virgin Islands. So, we still called it GENES but we had to keep going after the Empire State.

When they stopped funding corn and decided to reformulate the regions, some of them are pretty much the same. I think region four used to be Great Lakes but they picked up Kentucky, so they can't call themselves the Great Lakes anymore. This New England Genetics Consortium was NERC. It's got the same states as what it has now, just a different home and we lost Puerto and the Virgin Islands to region three which was the southeast region. Texas joined the mountain states, the heartlands. I think it's pretty much the same as it always was.

Kentucky must have left, I don't know NERC margin. I don't know where it came from. Anyway, so this is NYMAC, New York Mid-Atlantic Consortium and—so, that's sort of the regions.

One of our huge programs is the newborn screening programs and you probably pretty much aware of the new born screening programs. Every single infant in

the United States is tested for a certain life threatening conditions before the disease can cause permanent mental or physical retardation or death. All states, all developed nations and many of the developing countries have some newborn screening programs. The conditions are added to panel if they meet certain criteria. They're relative common in the population, though with these new expansion and with the (inaudible) that I'll mention briefly in a minute right below—relatively common. It's gotten very relative indeed. Some of the conditions are very, very, very rare. We found one or five in a literature or something. They're just—and there's pretty much a chemical thing. That they are detectable before clinical symptoms appear, some chemicals in their blood, in the babies blood is out of whack. And so, we can screen them out. There's available technology for mass screening, and there's effective treatment to ameliorate the effects of the condition, and that's really key.

Now, you know, Fragile X is something that is fairly easily identifiable, maybe in screening though, their working on it. But nobody really knows what to do with those kits, mostly young men but some women. Multiple--muscular dystrophy is another one where it's fairly easy to identify, but there's really, the treatment for this, mostly young men, has not progressed very far. So, there' really no reason to identify them based on the newborn screening line up here. Most of the conditions in newborn screening are genetic, two are mostly acquired during pregnancy, congenital hypothyroid, only about five to 10 percent of those are genetic and hearing deficits. It's unknown really. Some of them, they are few that

are genetically involved, but—and then we have a couple that are infectious, HIV and actually New York State is the only state that does test all of its newborns for HIV and toxoplasmosis. There's only a couple of states that do that, Massachusetts being one of them. There's a panel of core conditions or (inaudible) actually that has been recommended by the American College of Medical Genetics and this (inaudible), which is what some people have decided to call it. It's the advisory—it's the Secretary's Advisory Council on Hereditary Disease and Genetic Disorders in Newborns and Children.

Nobody could pronounce it 'til somebody just sort of strung together some syllables and it's easier than anything else. And the reason that we talk about—I'd rather talk about analyzing conditions is, these are just chemicals and we don't, they could be a number of different diseases. And we had this huge argument that five years ago is how many condition are in these core panel. Some people would count 46, some people would count 28 or, you know, depending on how many little sub-diseases you pick out. But the (inaudible) were all pretty well-defined. So, there's basically 29 (inaudible) and then that leads us probably about 45, 46 conditions. But there aren't a lot of states that do all of the core conditions. Those in red are really the only ones, at this point in time, mandate and do all the core conditions. Some of the states have the mandate but they haven't got the technology up. Some of them do it as requested or they do it unofficially. New Jersey, I thought they were up to speed but it's like, they do it but its not mandated. So, they don't get the red mark. And the green ones do all

the core metabolic or genetic, the laboratory conditions but they don't do hearing screening. Hearing screening is a bedside, a newborn, a nursery test. The way it's been reported is a little variable, but I can get that, let me get that.

UNKNOWN SPEAKER: Oh, thank you.

KATHARINE HARRIS: I'm going to get that. So, it's very variable. West Virginia is in our region and they only test for four or six, I know actually Pennsylvania tests for six, West Virginia only test for four conditions. West Virginia is working and they hope to be up pretty soon and NYMAC is really helping them.

Pennsylvania is a very strange situation, they don't have their own public health laboratory. And there's a few states that subcontract mostly to other public health laboratories for the newborn screening, Pennsylvania is an odd duck. It had a contract with pediatrics, which is for profit laboratory in Pennsylvania. The contract came up. It went out to bid and Massachusetts won the bid but only for their six core conditions. So, but all the hospitals, all the doctors in the hospital said, you know, we'll be crazy not to make sure our newborns didn't get screen for the core conditions. Somebody comes up with propionic acidemia, we can get sued because we knew they should be, the state didn't do it. It would be on our backs that we didn't do it. So, actually, 90% of the baby specimens go to, still go to pediatrics, where they get the core conditions plus the secondary conditions and Massachusetts is kind of annoyed with the whole thing.

There are some conditions that are universally screened in the United States. Phenylketonuria was the first condition at, that's what started newborn screening. All the credit goes to a doctor named Robert Guthrie, who had actually had a son who was mentally retarded not from PKU but he also had a niece who had PKU. Children who are not identified before they're symptomatic usually have gross mental deficits. They're very quite retarded around about 50, an IQ about 50 to 70 maybe. And he identified that, well, actually the treatment I think was identified in Switzerland, but he identified the test that you could identify PKU. He started with urine and fear at that point was that the laboratories might get 250,000 diapers which we didn't really like, but then he was able to develop these test on a blood spot, dried blood spot. So that started in the United States in the early 60's, New York started in '65. I think the first one was Massachusetts started in '64, maybe '63. I think certainly by '70, all of the states were doing PKU. Sickle cell disease is universally done. Yeah, that's very recent some of the states, I think Alaska added sickle cell disease. They figure it costs about \$4 million to diagnose one child with sickle cell disease just because their population is not at risk.

Sickle cell disease does tend to be a disease of African-Americans and Hispanic with African-American roots or other ethnicities and races around the malarial section of the world, the tropics. But it is universally screened. Congenital hypothyroid was added to New York State's panel in '78 and all of them added it around that time and galactecemia was also added to our panel in '68. And you

can see this incidences one child for PKU, one child in almost 19, 000 kids will be diagnosed with PKU in the United States. Sickle cell disease is very common and this is the whole population of the United States, one child in 1800. If you could look at more just the African-American population it's probably closer to one in 400. Congenital hypothyroid, at this point in time, the incidence is one of 3,000. I just published a paper a year ago, quite nine months ago that noted that congenital hypothyroid has tripled in New York State and doubled in the nation and we had a conference earlier this week at the CDC trying to figure out, is this real? And then how do we figure out what's going on?

Why are so many kids--so this is practically in the last five or six years went about 1,700. So, it's really big, almost not quite as prevalent as sickle cell disease but it's—it's a simple, it's such a simple treatment. You just gave them thyroxin, a pill a day and they are not—they don't develop (inaudible), they don't develop mental retardation, growth problems, behavioral problems. It's just, so it's just one of those miracle, even better than PKU. PKU is a tough diet to follow especially as the child gets older again and can't have meats and just has to stay with a special formula. Sickle cell disease, the treatment is effective but not entirely. They get penicillin prophylactically, so they don't develop sepsis. But they can still have strokes. So they have to do other things and they're developing, they seem more and more, they're developing more treatment strategies.

Galactocemia is another diet-based problem. It's a metabolic disease as is PKU. It's also very hard to manage because galactose is a sugar and it's found

universally, body makes some of it and yet almost any of it will poison the brain. So, these are interesting conditions. You can't cure any of them. Hypothyroid is the most amenable to treatment but the other sick kids are doing well, they're turning to adults which creates a new problem, but it's wonderful.

FEMALE: Question.

KATHARINE HARRIS: Yeah.

FEMALE: (Inaudible) published a paper (inaudible) increased number, is that because of screening? The more screenings...

KATHARINE HARRIS: No, its not. If you look at the number of kids who were screened compared to the numbers, the incidence has actually doubled or tripled

UNKNOWN SPEAKER: Okay, that's what I wanted to know.

KATHARINE HARRIS: Yeah, it's not the numbers. Yeah, and we don't know why. We've got some ideas and that's our next step is to try test some of this hypotheses, but, is it in the water? We don't know.

UNKNOWN SPEAKER: You have the (inaudible).

KATHARINE HARRIS: Yeah, no. Yep, yep, thank you. NYMAC's workgroup number two and in our first cycle, 2005 to 2007 all of our workgroups had numbers, which made it interesting, try to figure out who was, which number was doing what projects. But we were just developing our focuses anyway. So, workgroup number two was looking at access issues and their major project that we talked about was to identify the counties of residence of newborns, diagnosed by the newborn screening programs to locate the treatment centers, to determine which children live too far away from treatment centers to receive adequate treatment, and to develop strategies to make treatment more accessible. The mapping software, the mapping was primarily done by Susan Swire who was one of NYMAC's project coordinators and she was a coordinator in charge of workgroup two. When she went on maternity leave, (inaudible) helped finalize some of our maps. So, I've done some additional work on them and I think they're even better. And Susan with her husband Kevin chose to use Microsoft MapPoint 2006 with spatial point add-ins. So, that's the one I'm going to show you. And when I found, I just put this in as a poster and then they said they want me to do a skills building.

I'm going, I think I better learn how to use this. So, I did. It's not that difficult, but it's got some interesting things. So, our process was to collect the data from the eight state newborn screening programs in NYMAC. The cases of PKU, sickle

cell disease and congenital hypothyroid from 2001 to 2005. And we chose these three conditions because they're quite, they have a very high incidence.. We will be going to find a fair number of these children. Their universality in the newborn screening programs and representation of three of the four types of specialty care centers. As I said before, PKU and galactosemia, they're metabolic diseases. Congenital hypothyroid is an endocrinology. It's part of the endocrine system. And sickle cell disease is a hemoglobinopathy. And the other type of sickle cell disease is, respiratory or actually its pulmonary.

And that's for cystic fibrosis and a lot of the states are doing that. But it's not universally included in the state panels and our experience wasn't that broad, so we decided not to look at that section. Cystic fibrosis actually has a pretty good network for treatment because of the Cystic Fibrosis Foundation. So, we let them handle this and we look at the one that newborn screening really created. The only data that we collected on the newborns was the five-digit zip code and the diagnosis, so we don't know who these kids were. Then the data had to be validated to correct or remove invalid zip codes. We didn't have their addresses, so we couldn't. And I looked at the newborn screening data and it's always interesting. I think it's filled out by nurses, the moms, some of the moms don't want to tell you where they live. There are issues. So, they just make up zip codes that don't make any sense. If you have the rest of the address at least you can find a zip code that goes along with that if it's the real address. But there aren't too many of them. And sometimes for our purposes, it really didn't matter if

it was for Manhattan, for example, 10001 or 10026 and I'll show you why. But even with, there's 10,000 zip codes in the United States and probably three or 4,000 of them in our eight states.

So, there were no newborns diagnosed in some of the zip codes which caused a little bit of an issue. We had to learn how to deal with what. Hmm, let me see. Why is it doing that? I don't know. Let's see what we can do here. Okay. Not good. Let's see what we got here. I think I'm running into a—so, we're just going to have to—it's here. So, all right. We're back on track. I knew it was here. Ain't that weird? So, we might have to do it from here. All right, I'm sorry but let's see if we can make this a little bigger. I don't know why it won't show it. But you see enough of this from here to and you got the, there and you can get as close as you want obviously.

So, we decided to map the counties using the five-digit zip code. One of our issues was confidentiality and if theoretically if there's one kid in a zip code with a condition, you can track it. You could find out who that child was. And we didn't want that to be at all possible, so we decide to map counties using the five-digit zip codes and there were no diagnosed cases in a county, we found that we had to put in a dummy within acceptable limits for at least each zip code. And for a zip code in each county, so that we would get some of the result that we wanted. When we first did the mapping, we found some problems. So, we had to sort of put some dummy codes in there. So, for example, this is just a bunch of stuff

from New Jersey, apparently Bergen County and Essex County and Hudson County didn't have any PKU. PKU is relatively rare. So, we just had to put in a zip code from those counties and put in a zero as suppose to a one. So, we're going to—when you first open MapPoint, it comes up to this wonderful United States and this looks a lot like Google maps or any other mapping programs, and somebody has told me you can do this stuff with those software but I've never been able, never paid enough attention to them to find out if it really can since we're working with this one.

So now, but it's pretty easy to zoom in to the NYMAC region and you use—here, we go. So, here's a, for example, is when we zoomed into the NYMAC region and you can see, you know, New York State, Pennsylvania, West Virginia. We got Virginia down here. Unfortunately, we never been able to figure out how to get rid of the credits here, and then just go up the coast. So, that's what we get there with a NYMAC region. Actually, I can—let's see, we can if you want to see how this really works. Let me get to—I want to launch this, so if you got the time and the interest, I can launch it so we can see how it really works. I think I can. It says, do you want us to find you but it doesn't do that. Anyway, so this is, as I said, this is what happens when you open it up and you can use this box over here to zoom in on something, so if you click on that and then you sort of figure out how to draw a box around the region that you're interested in.

So, I have to finally learn how to draw one big enough to include all of New York City and Virginia. Then if you click on it, you'd zoom right to that area. It gets a little confusing with all that, all of the roads and stuff in there, so you can easily go to a data map which is there—oh, yeah, there's the states. We never figured out how to get rid of the other states that we weren't interested in or how to get rid of the big cities that had decided they wanted to show out, but that's all right. We managed to get around that. So, when you want to add the data to it, just go up to data and all of the Microsoft stuff has these wizards, so you go to mapping wizard, and we use this shaded areas. You could do other ways to demonstrate it. We'll use a different thing for showing the treatment centers, but basically we'll use the shaded areas. So, you can hit next and we want to import data.

So, you just click on that and do next and the one we were using, I'll use sickle cell because there's a lot more kids diagnosed with sickle cell or hypothyroid would be about the same. So, we'll use this one, this here and we want to use-- we've got the providers on one sheet and the sickle cell disease is on the other. So, we'll use the sickle cell diseases for the mapping. And it's interesting this comes up, and I don't know exactly how it decides but I saw this happened. We don't want it, it says it's going to match records for street address and we didn't want that. So, what you have to do, and it thinks those dates are street address, interesting.

So, we just tell that's other data and the way I learned to do this, whether it's the right way or not, is I don't know. That's the way that it works as one of those old, how did this happen. So now we're going to match records for zip code, so you push finish and it does all this cool stuff. It's looking at zip codes, and our zip codes, and counting up the numbers of one and zeroes, counting the ones, adding the ones basically. And we were looking at county, so it could do zip codes. But there are so few kids that even for sickle cell disease. And it's much more informative to look at the county, so we'll get the county for that one. It's just drawing push pins at the map. We decided to use discrete equal ranges and for some—you know, actually, as I was doing this over and over again, we always use five but it doesn't really matter what uses five because what were looking for was there's—that will help in some if you were looking at how many treatment centers do you need in an area.

But we are looking, for this purpose we wanted to know how many kids were outside of it, you know, where there was just one case. So, we can use three just as easily. And what we're going to do change this to zero. We want the first one, the first division to be zero because if there aren't any kids in that county, then we don't care. I mean, yeah, we know this is a snapshot and there could be kids in that county another time. But for now, we'll just stay with one. And then go up to here and we'll just take this one to five, say, because that's going to show us all the children from one to five. And we'll go up here and we can let everybody else be from six to 220 because that's—we don't really at that point about--that's

not what we're looking for this point. You get more colors. It's more interesting. But, so, here you can see all the white areas have no kids with sickle cell disease. The real dark green ones have more than five, but the lighter green ones are the ones we are really concern about. Those are the ones where there's one, at least one. The green areas in general we're concerned about, all the green areas, The white, we're not. So, then what you do, we want to add the treatment centers. So, now you're going to import data wizard and we use the same—let's see if that's right. We're going to do this and again we want to—we're going to match it for zip code and then found some things that I didn't like so we're just going to skip these because it's not really critical. We're going to use push pins because we like push pins. So, we're going to throw and we want it, we want them to be zip codes because we really want to know exactly where those kids are. Those zip codes are maybe in the county or whatever. This much, otherwise, you just take the center of the zip, of the county. So, center of the zip code to much smaller area.

And one of the things you may know about zip codes that I learned was with the three-digit zip codes, Manhattan is all three digit zip code. It's 100, all of the 100s are Manhattan. In the Bronx, it's all 104 and I can, I know zip codes in New York, I've been looking at them too long. But Upstate New York, if you go to 122, which is Albany, the Albany County area, there's also 122s way up north. They're outside counties, so when the post office was assigning zip codes, they start, I don't why, they started finding random ones. So, if you got the random one, it

really distorted what you were doing. So that's, so the three-digit zip codes, we felt that counties would work better. So, that's an interesting anomaly.

But for our treatment centers, we can just do this. Fine, whatever. Oh, I see what it's doing. Okay, oh, I think I know what happened. Let's go back. Is this--let's start over with this. See if I got this right. Sure. Yeah, (inaudible). Okay, we're going to start over but it won't take very long, I promise. And the edited providers in Manhattan, there's a lot of centers that do sickle cell disease. But we only, we only want to know that there's one, at least one. Again, more than one is just going to mess thing, confuse us and mess things up. So, that's okay. Excuse me. I went to the CDC and got sick.

So all these spots are where the treatment centers are. And we really, I don't like those spots, so we're going to change this. Why did we do that instead of--this is the fun part of when you do something like this. And, oh thank you so much. Let's try push pin, try that one instead or we're going to set the symbol. We can use some color. We'll use red; red will probably show up good. Okay, so now, I had to use red push pins and you can see, say, for West Virginia, there's one red push pin. In this western part of Virginia, there's one push pin. In upper New York State--looks like we somehow lost the very top of our New York State--oh, stop. Now, we lost Virginia. But that's, okay, well, that's okay we can deal with this right now. So anyways, you can see the kids up in say in Jefferson County might have to go a long way to get a Syracuse if they have sickle cell disease or the

kids in Erie County might have to go a long way to Pittsburgh. And that's one of the things we were concern about. So, one of the things we wanted to do, let's see, I might have to get back to my notes.

Let's go back to my notes and see how we do this, this is the demo, that's why it's good because--oh yeah, right, thank you. Okay, now I remember. We want to draw a radius, and down here on the draw bar, there's a cool radius here. So I'm going to take that and I'm going to put it on Pittsburgh. Now we had some debate, you know, how far should a family have to travel to get services and, you know, maybe 50 miles, 75 miles depending, and the (inaudible) said maybe 75 miles would be okay because those kids can be really seen by a primary provider there, treatment's not that complicated. Sickle cell, a lot of the primary providers don't want to do sickle cell because any time they get a cold, their sickle cell disease is impacted or there's whole sorts of other problems with that. So, you know, you can pick a number and 50 miles might be about an hour drive. So, let's say we're going to go here, and we're going to pull out a 50-mile give or take. Let's do 60 rather that, to me, we're can drive 60 miles an hour.

So, there's a circle around that area and then the next one for this one down here, draw another one here. So we, we ended up drawing all this 60 or 50 or whatever miles circles. And if you look, say, well let's look at just this area up here, we can do this for, so we got one in West Virginia, one Pittsburgh, one in Buffalo for example. You know, and those are about 60 miles. Okay, these guys

in Erie are outside of the circle. So these, any child with sickle cell disease and there may be one, there may be up to five.

UNKNOWN SPEAKER: Could it be possible for those people to have been on the board are actually going to Ohio for treatment?

KATHARINE HARRIS: Sure, absolutely. Even if they, right, I mean say there's one in Akron and theirs is, it's not in our region, so I can't. There are some issues going out of state, especially a lot of these kids with chronic conditions get put on Medicaid. Most of the states have a waiver that the parents didn't come for a kid with a chronic condition, the kid's income counts. Their parents' may not be considered to qualify them for Medicaid. But then they go out the state, does cause a problem. But some of them do, there's no question.

So, if we go to Akron, Ohio, that's one of the big city there, and there's still areas just, and we don't know that there's this treatment Erie. Pennsylvania, most of the states, most of the states do certify treatment centers and there are just not one in Pennsylvania in Erie. So, those kids up there are outside of that 50 miles. You get down the city and, you know, this whole area it's pretty good even, say, for Albany. If you draw that. Upstate New York is fairly ethnically uniform. So, you don't get a lot of sickle cell disease outside of the urban areas. So, you know, we don't really have to worry too much. There might be one or two. Jefferson County is interesting because there's a, there's a naval base there. Why there's a naval

base in Jefferson County, but there is. Actually it's for Fort Drum, it's military. It's army. There's a naval in Saratoga. So, parts of and I don't know where Fort Drum is in Jefferson County but as far as we know, there's no way, say, down in Broome County, if you're in Binghamton, there's no recognized center or if you're out here in whatever this county is, it's not marked. Does it say? It's a long way from anything that's got marked. We do think there's one somewhere in Hershey which is over here some place. But that's still outside of the 50 miles. So, the issue is same all the way around. It's going to be hard for these kids to get the services they need. In other ways, there's also just some of the fun stuff and we'll just show you one. You can create a drive's time zone. So, we want an hour, how many minutes we want? We didn't really look at that but, what? What?

UNKNOWN SPEAKER: No.

KATHARINE HARRIS: Because I didn't plug it in, I'm sorry. Turn it on.

UNKNOWN SPEAKER: So, Katharine, you taught yourself how to use that program?

KATHARINE HARRIS: Yeah, yeah. But I had done some mapping before.

UNKNOWN SPEAKER: (Inaudible) for you?

KATHARINE HARRIS: There's down there, yeah. Okay. Here, here, here. Oh, it's two-part (inaudible).

UNKNOWN SPEAKER: So, what could be (inaudible) the distance between the islands? And how would that be (inaudible), you know, the treatment centers on some of (inaudible).

UNKNOWN SPEAKER: No, that's not of the problem. Most of the treatment centers are on the urban areas.

UNKNOWN SPEAKER: Uh-huh.

UNKNOWN SPEAKER: And nearby island (inaudible) drive from one county to get...

UNKNOWN SPEAKER: Right.

UNKNOWN SPEAKER: You know, you got to fly over and that's a big expense. You know, just (inaudible). Usually there are some programs that (inaudible) some of it's...

UNKNOWN SPEAKER: Yeah, exactly.

UNKNOWN SPEAKER: Still, if you've been on call to—so, (inaudible) identify.

UNKNOWN SPEAKER: Yeah. Mm-hmm.

UNKNOWN SPEAKER: The (inaudible) reminds me she's also (inaudible).

UNKNOWN SPEAKER: Yeah.

UNKNOWN SPEAKER: You're so isolated, but you know you got to take these cases.

UNKNOWN SPEAKER: And are you finally familiar with the issues are (inaudible) sickle cell...

KATHARINE HARRIS: There are sickle cell (inaudible), there's four and they're all in Oahu because they're with the naval base.

UNKNOWN SPEAKER: Oh, okay.

KATHARINE HARRIS: That was--and the reason we know this is, there's some sickle cell money out there in Hawaii. Hawaii gets a lot of federal money there. They have some great grant writers. They come up with some great ideas. And they got awarded the sickle cell grant. And when we had--and I'm part of that

group. And so, we asked them how many they get, and said well four but they're at the naval base. So I was like, what? But, in...

UNKNOWN SPEAKER: You're, you're a transit passenger...

KATHARINE HARRIS: Yep.

UNKNOWN SPEAKER: And so, your transit possibility within an hour?

KATHARINE HARRIS: Yep.

UNKNOWN SPEAKER: So, you know, you might have four days you're in, (inaudible) one.

KATHARINE HARRIS: Or they'll have a different four. I mean, they'll the four kids who were there, born there and now shipped out and the other one, some other one, the kids are coming. So there's, that's a different issue but it's a very strong issue. They're still in Oahu, so it's fairly close to their medical center.

UNKNOWN SPEAKER: All the people in (inaudible).

KATHARINE HARRIS: Yeah.

UNKNOWN SPEAKER: It's more centralized (inaudible).

KATHERINE HARRIS: Right. Right. Right. Anyway, so let's draw a 60-mile drive time. I can draw a way around this area, and this is pretty fun. So, you know, a circle doesn't make a lot of sense because you don't know where the roads are and you write into Lake Erie or you go out to out into Atlantic Ocean. This one is really--let me make this a little narrower. I'll make it just this two. So, but this is really more real and that's put a drive time and I'll show you how this works with this one in Maryland. No, oh, sorry. So, put a drive time zone around, around this one in Baltimore.

So you can see that, let me make it a little narrower, so it's not quite as overwhelming (inaudible). It doesn't pretend like, maybe there's a bridge there and so it goes across the bridge. But it's more rational, I think, than the circles. It's a little harder to read but I think it's more real. So, you can decide on that. And this is, it's going to take you 60 minutes, it's going to take an hour to get your kid in. And if you're doing this--for hypothyroid, again, those kids are very stable, primary provider can work with them, especially once they're a year old in there. I mean, the first year, their growth spurts are all over the place. And they may need to get to the center more often. But once these kids are older, they may need to go to the center once a year. At that point, it's not as much of a hardship certainly. Still a hardship for a parent to take a day off which is always can be, you know, depending on where you work, I work for the state. I can take a day

off. It's not a big issue. But if you're working for hourly wage at a store or something, you may not have sick time or you could, maybe Ellen, you've used it all up. So, you have to, you might have to use some much more valuable time. So, I think I like this drive time radii because it's kind of real. But, so anyway, so this is the program. It's quite easy. I mean, I've made some fix and starts, but it's not that hard to learn. And it's, you know, that you buy it and there are several other (inaudible) used map professional and get similar results. But I think it really gives you a good idea of where the issues, where the problems are. If we were to go out to Hawaii, you could, drive time would be much different. You'd have to stop. But I, you know, that's pretty much it and I would be interested now, now what do you do with this stuff? And I can to that, those slides now.

So, this is a new cycle, 2007 to 2012 (inaudible) gone to five year cycles. And we now have named our workgroup, so we have a distance strategies workgroup and they're developing distance strategies to help families access specialty care services over longer distances. So, one of the things and we've started out calling this group, telemedicine. But we didn't want to be limited to telemedicine, so we go wait for the distance strategies, but we really are focusing at first on telemedicine which—can, you know, the sort of idealized, this is videoconferencing. You've got a child and a family, and a primary care provider or a nurse or somebody out in the rural area or something, and then you have the same capabilities with a specialist in the medical center. And they, so you can focus up the camera technology. It's over the Internet, it's just amazing what

you can see and they're able to look at the faces, especially when these kids don't look any different. We've said, you don't really need telemedicine to do PKU. You're not looking, I mean, other than I think if you're talking to the family you do need to look at them in the eye and be able to relate somehow and see that they're paying attention, body language has a lot to do with.

When you're examining a child, you don't need to examine a child for PKU necessarily because they don't look different. But you do have these unfortunately called "funny looking kids." Children who don't look right and the genetic--clinical geneticists are trained to look at different things, how wide are the eyes, where are the ears placed, you know, other you know look at the hands or the feet or other parts of the body. And they can try to put these all together into a syndrome or something new. Parents want to know is, if there is a syndrome, they would like a name, most parents want a name to what's wrong with their child for a couple of reasons. By having it, giving it a name, you can go back and look at other children. If they had that name and what's happened to them, and what has medicine done for these children, what can I expect? And it's always going to be variable, but at least you have an idea of what to expect.

And then the other thing to have a name is just, you know it's something. I mean, and we've all had these weird elements that don't mean anything. They don't add up to something, but they drive you crazy because you don't know what it is. And if it goes away, great. You can forget about it 'til it comes back. But for the

newborn screening, in most cases, you don't need video necessarily, especially on ongoing basis, the physician—and we'll talk a little bit about that.

But let's look at telemedicine first, so those who are health professionals discussing a case over the telephone. That's that simple. Just a consultation and that's been happening for hundreds of years or maybe not. How long is a phone have been out? Hundred years. Before they used to send snail mail. But it's been happening as long as there've been telephones. Talking to colleagues, I don't know what to do this child, especially among, among specialists or using the satellite technology and videoconferencing equipment for a real-time consultation between medical specialists in two locations. So, between a medical specialist and a primary provider, genetic counselor and somebody at the other end who is helping the family understand what's being said. And apparently they're doing a lot of videoconferencing out in Hawaii and in the western states because the distances are so long and it is so hard to get a—and it's working out very well.

They found, you know, having somebody , a professional at each end to talk to the family and really try to read what the family is going through and comprehending do they need to back up. People are, especially if you can't see them, they're not, do you have any questions? Who has question? I mean, they may not have a clue what you just said. And they don't know how to ask the question because they don't even know where to start. So, by looking at them, you might be able to say, you know, what do you think I just said? So, it's much

easier to interact with the family in the video. So, this is how New York State in, I'm showing you, in Pennsylvania, but New York State has implemented the new hub-spoke system. They're using it for rurals, for strokes in rural hospitals.

They've got a radiologist who can read the MRI from the stroke victim, and work with the primary physician at the emergency room in a rural hospital. So, they don't have to transport the patient as much they can do the procedure and then read it at the specialty care center then talk back and forth.

So, you know, you can have a spoke out in Erie, you can have one, stop it, I didn't do that. You have one in state college, you can have one over this way, and they could all be talking to the people in Pittsburgh, for example. So, that's the hub-spoke system. And we're trying to get that put in place for genetics and outpatient in New York State. They only put it for inpatients for emergency rooms, we're saying this is really something. And the deal that they put it together for was for Medicaid. You could get some reimbursement for it. So, we're trying to get them to expand that to outpatient where it's really crucial.

Another, stop that. So, another possibility would be, we do this for PKU, you can do it with congenial hypothyroid. Do this field blood spot specimens, where you monitor somebody with PKU or with hypothyroid as you can take a little bit of blood, you can put it on a dry blood spot, you don't even have to take a vial, just prick their finger there once they get old enough, send this into the newborn screening laboratory and then report it back to the specialist who will call the

primary care doc. So, you got somebody up here in wherever this is, I can't see what it is, some place up in northern New York State, outside the Adirondacks who does this field specimen, sends it to the newborn screening laboratory in Albany who sends the report back to the specialty care center at upstate in Syracuse who then talks, this is just, could be a conversation, telephone conversations. So, this is the level, the phenylalanine, say, is stable. This is good or it's not good. You know, what's going on with the diet kind of thing. So, they can do this kind of--you don't need the high-tech stuff, you just draw the blood and use the phone, send the mail systems. So, it doesn't have to be crazy, it could be quite simple, some of these strategies. Or another one, somebody mentioned is, just have an outreach laboratory or something. Hi.

Anyway, you could have, you know just an outreach. Those newborns have a high immunoreactive trypsinogen which is indicative of cystic fibrosis and then most of the lab will reflect any of these high IRTs and do a DNA mutation analysis. If they find one of the, most of the panels only run about 30, 40 of the mutations because cystic fibrosis has so many mutations. But if they didn't find one of them, you don't know that because the cystic fibrosis gene is so fragile, you could have a different one that you haven't identified. So, you really don't know at that point, is that person a carrier of, say, delta 508 or do they have another mutation that's going to cause a disease.

A carrier usually doesn't have any symptoms, he's really basically healthy. Most of our bodies are, our body systems are so redundant, you can live on a little less of this cystic fibrosis factor. You can do just fine, but if you have two, then you're going to have the nutritional problems, the respiratory problems. So, we don't know. So, most of the newborn screening programs who do screen for cystic fibrosis, for example, require that the child have a sweat test, which is a little tricky to do from what I've done them.

I'm a licensed medical technologist. As a matter of fact, I've done of them, and I haven't any clue what I was doing, so I wouldn't rely on mine. So, mostly we want this done by people who know what they're doing. But if it's hard to get there, you know, you can at least have it, so you can send out from the medical center, you can send a technician. We've got somebody out there. So, that's one person who is being paid to do this. It's not the mom or the dad who is losing a day's work to take their child in. Somebody's going to go out there and pick, collect the specimen or maybe a couple if you're lucky.

So, the tech is going to travel from Charleston, West Virginia down to Meridian, I think that is in West Virginia, go down there, collect the specimen, bring it back up, they'll analyze and then they'll call the primary physician. So, that's another outreach low-tech, outreach distance strategy that's a possibility. So, that's pretty much it and you may have some other ideas of strategies or ways to use this kind of mapping software.

UNKNOWN SPEAKER: (Inaudible) software, how do you choose the Microsoft product compared to other mapping softwares?

KATHARINE HARRIS: I think it's really, well I don't, this was done because Susan Swire's husband does this stuff and he liked that program. It really is like that simple. There's other stuff and as I, as I said, somebody told me you could do that stuff with Google maps, and Map Quest. I didn't take the time to learn because I had the software to use and we'd already done it, so I knew we could do this for this application. I think they all have very good qualities and I wouldn't be in a position to know which was better or worse. If you have somebody who knows what they're doing, they probably already have one that they like, and they've learned how to use it so they would use that one. But if you're fresh and clean, this one was, it's fairly easy to learn. We did have cheat sheet that Susan had developed, Susan and her husband had developed. So, when I was learning how to do it, I didn't have to go through the tutorial or something but they're pretty self-explanatory.

UNKNOWN SPEAKER: You used zip codes. Was there any zip codes based on the mailing address or...

KATHARINE HARRIS: Their residence?

UNKNOWN SPEAKER: (Inaudible).

KATHARINE HARRIS: Yeah, residence.

UNKNOWN SPEAKER: So, you didn't have problems with people using the P.O. Box.

KATHARINE HARRIS: Yeah. Even if we did, I think for our purposes, it probably didn't matter. I mean...

UNKNOWN SPEAKER: (Inaudible).

KATHARINE HARRIS: Yeah, yeah. I wouldn't go that far to get their mail probably. So, but I think with the newborn screening—oh, I've seen some POs, post office boxes.

UNKNOWN SPEAKER: Do (inaudible) to ask PO Box?

KATHARINE HARRIS: Sure, sure. Yeah, yeah.

UNKNOWN SPEAKER: ...usually have our newborns here in screen data and (inaudible).

KATHARINE HARRIS: Love you.

UNKNOWN SPEAKER: So...

KATHARINE HARRIS: I wish we can do that.

UNKNOWN SPEAKER: So, national zip code.

KATHARINE HARRIS: Mm-hmm, yeah.

UNKNOWN SPEAKER: So, that's where we're looking at?

KATHARINE HARRIS: Oh, yeah. Oh, yeah. I maintained that parents are going to be--most parents do newborn screening, very few parents refuse newborn screening and almost--there's only two states that have, that require informed consent. Almost all the other states require informed descent. You can opt out, but most in 99 point, who knows, we only have a couple a year that may opt out, mostly for religious reasons. I don't know, most the doc say, no, don't do that, you know. Yes, but they're going to say, there's never been a problem with my family.

As I said before, all except for hypothyroid and the infectious diseases, they're all genetic, they're all recessive. So, you don't see them, especially in our family sizes. Well, Pennsylvania is interesting with PKU, and if we've match PKU, the

Amish has a founder gene for, not for PKU, for MSUD, for Maple Syrup Urine Disease. And Pennsylvania didn't, I think, now does test for MSUD but we used to have a lot of families that would drive to Binghamton because they knew New York State would screen for MSUD, they wanted, they wanted their child screened for that because it's a devastating condition. And it's very, it's very common in that population because they're so intermarried, so very small contained community.

The (inaudible) Jewish are the classic ethnic group for recessive diseases. They just, the least goes on and on and on because for so long, they were tied up in their own little communities, excluded from everybody else. So, for a parent to say that we don't have any of those things in my family, they need education to say, yes, you're probably right. But if you're wrong, the cost is so enormous that you don't want to have to go there. And what is it? It's a stick in the heel. You know, the kid, yeah, the kid will cry, but the kid's going to cry anyway. So, go for it. So, we have very few people who would opt out.

UNKNOWN SPEAKER: In the pore screen, the recommended (inaudible).

KATHARINE HARRIS: Yeah.

UNKNOWN SPEAKER: ...point sticks.

KATHARINE HARRIS: 29 analytes, yeah. Though actually that, that's not true. I think they count like three or four hemoglobinopathies. So, it's probably is like, they say 29 conditions but it's probably that 25 analytes.

UNKNOWN SPEAKER: Now, there's new finding that is going to help states move up to that board.

KATHARINE HARRIS: There's really no federal funding that I know of but...

UNKNOWN SPEAKER: I think there's going to be...

KATHARINE HARRIS: Is there? Okay.

UNKNOWN SPEAKER: Something that's going to facilitate states moving to at least be able to screen for that or (inaudible).

KATHARINE HARRIS: Yep.

UNKNOWN SPEAKER: (Inaudible) MCPs were talking about it.

KATHARINE HARRIS: Yep.