Centers for Disease Control and Prevention
EARLY HEARING DETECTION AND INTERVENTION
Ad Hoc Group - Teleconference Agenda
September 4, 2001

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LEE ANN RAMSEY: I want to welcome everyone. My name is Lee Ann Ramsey with the Centers for Disease Control and Prevention, and this is our September teleconference on Early Hearing Detection and Intervention. Thank you very much for joining us. Please remember to mute your phone when you're not talking. Today's conference is being recorded and the transcript will be available on the Internet.

Before we begin our scheduled program, are there any announcements or comments from any of the participants? Anything?
(NO RESPONSE)

LEE ANN RAMSEY: Okay. Well, then we will skip the Federal Update. I believe that Irene Forsman and June Holstrum are in a meeting. So they may be joining us just a few minutes late. But Dr. Jean Johnson in Hawaii is on the line with us, and she's going to be addressing a study that they are doing on the efficacy of OAE and AABR in identifying hearing loss. I'll turn the program over to you.

JEAN JOHNSON: Okay. I'm reporting on a research proposal that was developed about a year ago. The proposal was in response to a request that originating from the Centers for Disease Control and issued by the Association of Teachers of Preventive Medicine. The request was in response to the concern that as more and more hospitals implement Universal Newborn Hearing Screening concerns have arisen about the accuracy of the two-stage protocol in identifying children and whether perhaps some babies with significant hearing loss were being missed.

This research design looked at the OAE/ABR protocol and whether or not some babies who had failed the OAE but passed the ABR might indeed have a significant hearing loss.

The research study was designed to address this question. Our purpose was to define a birth population of about 30,000 babies from which a diverse sample of about 1,500 infants would be studied. These are the infants who failed the OAE but who passed the ABR. We are working with 14 major birthing centers that have been using this two-step protocol with considerable success. We established criteria for screening levels, for pass/fail referral rates, for thoroughness of follow-up at those facilities. The facilities that in the study include: Arnold Palmer Hospital in Orlando, Florida; Huntington Hospital, Plainview, Long Island Jewish Medical Center, and North Shore University Hospital in New York; Kapi`olani Medical Center here in Hawaii; Good Samaritan Hospital in Cincinnati, Ohio; Lawrence and Memorial in Connecticut; Via Christi Regional Medical Center in Kansas; Jacobi Medical Center and North Bronx Hospital in New York; and Women and Infants in Rhode Island.

The sample will be representative of the nation's ethnic and racial births. We began enrollment on May 1st of this year. At the end of three months of enrollment, we had enrolled 255 babies. We will begin doing the audiological assessment of those babies when they are about seven months of age.

Other people involved in the research project include: Karl White of Utah, who is the research coordinator; Judy Widen from Kansas, who is overseeing the diagnostic audiological follow-up protocol; Yusnita Wierather is the project coordinator here in Hawaii; and I'm serving as the PI for the project. We should conclude enrollment sometime late in the spring. We hope to be able to begin reporting results sometime late next summer. This project is scheduled to end in September of 2002. Are there questions?

JACKIE CUNDALL: This is Jackie in Tennessee. Are you planning to re-screen them at different periods of time, or how are you doing that?

JEAN JOHNSON: No, we will not be re-screening them. At seven months of age we'll be doing actually an audiological diagnostic evaluation on each of them.

VICKIE ANDERSON: Question in Minnesota. This is Vickie Anderson. Will you be using insert earphones rather than a VRA paradigm, or how would you be doing that diagnostic?

JEAN JOHNSON: Judy Widen is overseeing that protocol. The protocol we will be to what was used in the NIH study. We will be using VRA and insert earphones.

VICKIE ANDERSON: Thank you.

LEE ANN RAMSEY: Any other questions for Dr. Johnson? (NO RESPONSE)
Thank you very much for your presentation. I believe we have Irene Forsman and June Holstrum on the line, if we could get a Federal Update from Irene at HRSA.

IRENE FORSMAN: In the past week, we've advised nine states that they've been recommended for approval, and those states are: Washington, Wyoming, Maine, Connecticut, Delaware, Maryland, the Northern Mariannas, Arkansas, and West Virginia.

We've also entered into a three-way interagency agreement involving CDC, HRSA, and the American Academy of Pediatrics -- to work more closely with state Academy chapter chairs who are then to engage their community partners in the newborn hearing screening initiative. As most of you know, we're anticipating holding our first grantee meeting September 24th through 26th, and that's a combined CDC/HRSA effort. As of last Wednesday, we had 21 pediatricians registered because of their affiliation with chapter chairs. I suspect we'll have a few more. That's about all I can tell you at the moment. Are there questions?

PENNY HATCHER: June, this is Penny. Just a quick question. Did AAP notify those state chapter people about the conference? How did they find out about it?

IRENE FORSMAN: The Academy notified the chapter chairs, twice, in fact.

PENNY HATCHER: Okay.

IRENE FORSMAN: It would not be a bad idea at all for you to talk with your chapter chairs if you know who they are. If you don't, you can get a list from the Academy.

PENNY HATCHER: Okay. I bet I could go on the website and --

IRENE FORSMAN: I'm sure you can.

PENNY HATCHER: Okay. Thanks for that tip, Irene.

LEE ANN RAMSEY: Okay. June?
J
UNE HOLSTRUM: I wanted to congratulate our 15 newly-funded CDC cooperative agreement states, and those states are: Alaska, Arizona, Delaware, Illinois, Kentucky, Louisiana, Missouri, New Hampshire, North Carolina, Rhode Island, South Dakota, Tennessee, Wisconsin, Wyoming, and the Virgin Islands. These states will join the 15 states that were funded last year in developing and enhancing their EHDI state tracking systems.

Over the past few months, several of us from the CDC staff have been visiting the first year funded states, and we've been very impressed with the progress they are making. Particularly impressive is the dedication and the enthusiasm of these states. As you're probably aware, we're not giving a whole lot of money out, certainly not enough money to establish a whole system. So, we're very pleased with the eagerness and the enthusiasm of these states.

Another thing the states have done in this last year is develop a list of seven national goals for the tracking and surveillance systems. They are developing performance measures for each of these goals in order to evaluate the progress they're making towards obtaining the goals. The document containing the goals and the objectives will be available for all states.

We also have funded a number of research projects designed to enhance the EHDI programs. One of those you just heard about from Jean Johnson, about the efficacy of the OAE/AABR protocol in identifying hearing loss.

Additionally, we have four states that are investigating genetic and other causes of hearing loss. This project started out with Utah in the first year and they'll be joined by Hawaii and Rhode Island and, through CDC, Georgia. Utah is also implementing a cost study of EHDI programs. The PI's for the cost study are Linda Goetze and Karl White. Colorado is looking into the integration of EHDI with newborn blood spot screening. The PI's there are Bill Letson and Vickie Thomson. Hawaii has just been funded to look at a project related to the outcome of children with hearing loss at one, two, and three years of age. Beppie Shapiro is the PI there. And Rhode Island will be initiating a study on psychological and family issues. Betty Vohr is the key contact there. As these projects progress, we will provide more information to you all on how they're coming on the outcomes. For more information on any of these projects, you can contact the various PI's or we (CDC staff) can give you that information. So that's the CDC update. Do you have any questions?

PAT DEWEY: June, this is Pat from Virginia. How many new grantees did you just announce?

JUNE HOLSTRUM: Fifteen.

PAT DEWEY: Thanks.

John Eichwald: I just wanted to point out that the principal investigator on our genetic study is John Carey.

LEE ANN RAMSEY: Okay. Any other questions and comments before we move on? (NO RESPONSE) Okay. Are Sandra Gabbard and Vickie Thomson on the line?

VICKIE THOMSON: Yes, we are. This is Vickie.

LEE ANN RAMSEY: Thank you. If you want to give the AAA update, that would be great.

VICKIE THOMSON: Okay. Both Sandy and I are here.

One of the things that AAA is working on is developing their website that will link (inaudible) out there but also really appeal to the audiologists professionally as well as the consumers, and we're also developing brochures. We have a series of brochures that we're in the process of trying to come up with, one that is for parents and a separate one for professionals. And we are going to be working with the American Academy of Pediatrics and Family Practice Associations to get their input on these two brochures. We'll also be developing an assessment brochure for families so they will know what to expect once they've been referred for their testing.

And then the last brochure that we're looking at is a combination of an early intervention hearing aid brochure. That will talk about different communication options, kind of describe those real briefly, and then also talk about hearing aids and communication.

I'm going to let Sandy talk about some of the other things that AAA is working on.

SANDRA GABBARD: Many of you may be aware that AAA is working on guidelines for audiologists or benchmark guidelines in the area of amplification. We chose this primarily because ASHA is in the process, or maybe actually has completed, updating their assessment guidelines for birth to three, and we felt there was a gap there in that practice for amplifications. Alison Grimes is leading that effort, and I do expect that we'll have something out fairly soon, certainly by the AAA convention with those guidelines.

The other thing AAA is doing -- and many of you probably have already heard about this -- is they're having educational seminars, and the first one (inaudible) session to our convention on follow-up from newborn screening and what comes next. It was very well-attended in San Diego. It's two days' worth of meetings for audiologists to give them information about diagnostic assessment and amplification intervention. The next one will be in Atlanta, Georgia, and there's information on the AAA website about how to register for that and the exact dates.

They're also planning one for Chicago and it's not up on the website yet, but I know it's -- they're looking at January dates. And Lisa Hunter is the one who is coordinating, I believe, setting up

these conventions. So if you're interested in finding out if they're planning to do any more, she would be the contact person for that.

I also want to use this chance to get a plug in. The Marion Downs National Center will again be sponsoring the National Symposium for Hearing in Infants which will be next summer. The dates are August 1st through 3rd in Breckenridge, Colorado, with a focus on diagnostic follow-up and early intervention on following newborn hearing screening. And if you want to get on the mailing list for that, feel free to let myself or Vickie know about that. Any questions?

UNIDENTIFIED SPEAKER: What's the timing of the brochures that you mentioned, Vickie, like the what to expect with the diagnostic evaluation and the early intervention hearing (inaudible)?

VICKIE THOMSON: That's ready to go to the board, again by AAA, in the spring. There are so many states that have already great brochures in place that we don't want to reinvent the wheel. So I've been just kind of slowly getting brochures from other states to look at, getting their okay to plagiarize whatever we see. California is a perfect example of a state that has just a whole series of brochures. So we want to kind of pull those together, and then they'll actually be for sale from AAA. That way you can put your own state information on them.

UNIDENTIFIED SPEAKER: Vickie?

VICKIE THOMSON: Yes.

UNIDENTIFIED SPEAKER: Question, will the brochures be available in more than one language?

VICKIE THOMSON: Well, we're hoping to do them at least in Spanish and then we'll probably have to go from there. It kind of will depend on the funding from AAA.
Any more questions? (NO RESPONSE)

LEE ANN RAMSEY: Thanks to both of you. I would now like to welcome Dr. Walter Nance of the American College of Medical Genetics at the University of Virginia. Are you there, Dr. Nance?

WALTER NANCE: It's a great pleasure to participate in this phone conversation. Let me introduce myself, first of all. I'm Dr. Walter Nance. I'm Chairman of the Department of Human Genetics at the Medical College of Virginia in Richmond. I've had a long-standing interest in genetic deafness and was chairman of a subcommittee of the American College of Medical Genetics that strongly endorsed the views expressed by the Joint Committee on Infant Hearing, which suggested that geneticists should be an integral part of the team that evaluates newly diagnosed children with hearing loss. I think the reason for that is our growing awareness that genetic factors play such a major role in the etiology of hearing loss. It's of interest to me that since the newborn hearing screening program was begun in our state, we've tried very hard to get audiologists to refer patients for genetic evaluation, and I think that one of the reasons why this

perhaps has not been as successful as it might be is the misunderstanding of the major purpose of a genetic evaluation. That purpose is to establish an etiological diagnosis.

There are well over 100 different genes for deafness that have been identified. Some of these are quite rare. Some of them are associated with other clinical findings. Some of those clinical findings can be of great health importance to deaf children and their families. On the other hand, some of the genes for deafness are extremely common and can be tested for with DNA diagnostic tests.

So we actually believe that all children with a confirmed hearing loss ought to be seen by a geneticist to help establish an etiologic diagnosis of these children.

In July of this year, I also was a member of a committee that was sponsored by HRSA and endorsed by the American College of Medical Genetics which drew up guidelines for the genetic evaluation of newly diagnosed children with hearing loss. Those guidelines should be available in draft form by the 25th of this month and I think will be presented at an upcoming HRSA conference. Basically, they spell out in some detail the kinds of things that a genetic evaluation should include and the expected outcomes of this kind of evaluation in terms of making a diagnosis and counseling the parents about the options available for them and their children.

One of the important areas that's had an enormous impact on this field is the tremendous progress that has been made in sequencing the human genome. And as I mentioned earlier, there are several hundred genes that can cause deafness. Well over 100 have been mapped, over 50 have been cloned and identified and, in principle, can be screened for. Our knowledge about these entities is rapidly evolving and I think that it is very important if testing is done for any of these conditions, that it be implemented by professionals who are aware of the limitations of genetic diagnostic testing and able to provide a reliable interpretation of the test results for the parents.

I am extremely excited by the coincidence of progress in our understanding of genetic deafness and the establishment of newborn hearing screening. Both events matured at exactly the right time for them to be mutually supportive. I think that one of the things that we can anticipate in the near future is the use of blood spots obtained in newborn metabolic screening to test for selected forms of genetic deafness. We know for example that all forms of genetic deafness are not necessarily manifested at birth and, indeed, not all forms of environmental deafness such as cytomegalovirus inclusion body disease are manifested at birth. So that a logical target for molecular genetic testing in the newborn period would be to try and identify those forms of deafness that might be missed in a newborn audiologic screening program and in this way identify yet another group of high-risk infants that need to be followed by audiologists to be sure that they don't develop a hearing loss early in infancy.

These were some of the considerations that were discussed at the meeting that I told you about in July. There were about 20 people at that meeting from really all over the country who helped put together these guidelines which should be forthcoming before the end of the month.
I would be happy to respond to any questions that people have.

UNIDENTIFIED SPEAKER: Could you clarify again where they will be made available?

WALTER NANCE: My understanding is that there is a meeting of HRSA, and I'm not certain exactly what conference that is, but it's going to be on the 25th of this month.

IRENE FORSMAN: Dr. Nance , this is Irene Forsman. It's the CDC/HRSA grantee meeting.

WALTER NANCE: Okay. And Mike Watson from the Colleges is going to present the recommendations at that meeting.

VICKIE ANDERSON: Question from Minnesota. This is Vickie Anderson. You mentioned that you would encourage audiologists to recommend genetic evaluation, and I do work clinically as an audiologist and I would have to say that probably at least 80 percent of the time, if not more, my patients' parents decline.

WALTER NANCE: Yes.

VICKIE ANDERSON: I always recommend it. And they were talking earlier about developing brochures.

WALTER NANCE: Yes.

VICKIE ANDERSON: What I think might be helpful would be a very parent-friendly brochure that gives some credibility to my inept description of what that evaluation would be that they could understand.

WALTER NANCE: Yes. I think that's an excellent idea. And when I have talked to other audiologists, as I've said, who have had exactly the same experience of recommending a genetic evaluation and then having a small percentage of people uptake on it, the point I've tried to make to them is that if they told the parents what I have done is to simply confirm the presence of a hearing loss, now that we know there is a hearing loss, we need to find out its cause. The genetic evaluation should be presented primarily as an attempt to establish the etiology and to find out what particular genetic environmental factor is causing the problem more parents would be anxious to have a genetic evaluation. There may be other associated findings that are of great importance that could emerge from this type of evaluation and establishing a definite cause can have great value for the child, the parents and other relatives. But I share the view that you expressed that for many parents who are in the middle of the grieving process and trying to find their way in terms of what to do with their child, getting genetic counseling may not seem to be an important priority. I think if this were presented as a diagnostic etiologic evaluation that, hopefully, we would have greater uptake on referrals.

VICKIE ANDERSON: Thank you.

UNIDENTIFIED SPEAKER: This is another person in Minnesota.

WALTER NANCE: Yes.

VICKIE Anderson: We actually heard from some parents that posed a question to parents we had in a training about, you know, having a genetic evaluation. And at least what I heard is -- and I've heard this before in my profession as a pediatric nurse -- that a genetic evaluation connotes family planning, you know, tell me, you know, what will happen if I have more children.

WALTER NANCE: Yes.

UNIDENTIFIED SPEAKER: I suspect it's not -- your comment of making it -- thinking of it in the context of an additional diagnostic evaluation I think is important, but I also think that there's a community norm and a connotation out there with genetic evaluation related to the next baby, or should I have one and what's going to happen.

WALTER NANCE: Exactly. And I think these are attitudes that we need to try to defuse, specifically with respect to evaluation of children with newborn hearing. I mean, a couple might say, well, we're not going to have any more children and we don't need genetic counseling, and I think that's a very common point of view. If this could be presented as a diagnostic procedure that would help us find out whether there are any other health problems that you or your baby needs to be concerned about, that's where -- I think we might get a greater uptake on referrals for genetic evaluation.

LEE ANN RAMSEY: Any more comments, questions for Dr. Nance?
HALLIE MORROW: This is Hallie Morrow from California. Actually, my question isn't for Dr. Nance. It's for Irene Forsman. I was hoping that, Irene, you would be able to forward a copy of these guidelines for the genetic evaluation of these children out to the state coordinators because we all can't make it to the meeting this month.

IRENE FORSMAN: I can do that, Hallie.

LEE ANN RAMSEY: Okay. That concludes our program for today. Before we close, are there any last comments, questions, or announcements from our listeners?

PENNY HATCHER: This is Penny in Minnesota again. I want to just pick up for a moment again, Dr. Nance. Has anybody, and also the listeners -- looking at some focus group activities with families, those that have used genetic evaluation and those that haven't (inaudible), how can we speak a language that is nonthreatening that opens it up? And then I was also thinking, Vickie and Sandra, what AAA is doing with the brochures and (inaudible) planning on one brochure related to early intervention and hearing aid. And I question there, are you going to have a genetic evaluation and how can we word it? What can the rest of us learn, because we're all moving that way very quickly.

WALTER NANCE: I think these are all excellent ideas that we really need to work on, because I am somewhat dismayed at the low uptake on genetic referral. For the first time, genetics really has something to offer parents and children with hearing loss because, in so many cases, we can make a specific etiologic diagnosis either by the clinical evaluation or by molecular genetic testing or both. Ten or 15 years ago, the genetic counseling of a couple with a deaf child was

often sort of hand-waving process about the possibilities that it was either genetic or environmental but now with the array of specific tests that are available, we can often make a very accurate diagnosis of what the cause of hearing loss is. But I think you're right, that we need to look at strategies to try to do a better job of advertising the value of this procedure.

IRENE FORSMAN: Penny, this is Irene. I think that the chances of getting -- or engaging families in the genetic diagnosis might be much better if the primary care physician or the medical home was engaged in that discussion. They may very well not be.

UNIDENTIFIED SPEAKER: There you go with that -- Didn't I just hear you've got, working with the state chapter chairs, HRSA, CDC, and AAP. Maybe we can, meaning here in Minnesota, get our state chapter chair to look at it.

WALTER NANCE: Yes, this is Walter Nance again. I have heard it said that the way that newborn metabolic screening got off the ground and is the tremendous success that it is today was to obtain the active support of primary care physicians in the pediatrics profession and get them committed to the idea that those tests should be done and followed up on. And maybe this is the best route to go in terms of increasing the utilization of genetic evaluation and counseling services for these children.
YUSNITA WIERATHER: Dr. Nance, this is Yusnita Wierather from Hawaii. I really strongly support what Irene has said because of our (inaudible). We send the families requested to genetic evaluation, but the (inaudible) to even know where to start and what to do next. And the parents feel like, what should I do, especially for the states that do not have geneticists.

WALTER NANCE Well, most of the states of the Union have genetic disease programs that were started in the '80's by federal funding. So I would be surprised if there's not some resources available for the state that patients could be referred to. There are many other sources of information about where geneticists are located that they could utilize to identify appropriate places to refer them. For example, the American College of Medical Genetics maintains a roster of Board Certified Geneticists and the website Gene Clinics (www.geneclinics.org) contains a geographical listing of sites at which genetic evaluation and counseling can be obtained.

VICKIE THOMSON: Hi. This is Vickie Thomson. I just want to share with everyone that we have asked the geneticists on our advisory committee (inaudible) to come up with just a little letter that describes a genetic counselling session or describes what (inaudible) that the geneticists will be doing (inaudible) share that with the medical home and with the audiologists to get to their families. But along those same lines, I think it would be great if the AAP and Dr. Nance or the American Academy of Genetics could come up with a really family-friendly brochure that would be related just to infant hearing that we could share with our medical homes and audiologists.

UNIDENTIFIED SPEAKER: I agree. I think that's a great idea, Vickie.

WALTER NANCE: I think that our group, when we met to consider this, felt that there are so few geneticists in the country that children should not be referred for genetic evaluation of hearing loss until after the presence of a hearing loss has been confirmed. There are certainly

forms of genetic deafness in which establishing a definite diagnosis can have an impact on the management of the child. I'm thinking of things like Usher Syndrome where you're dealing with progressive visual loss as well as hearing loss. Another issue is the fact that now that we have the ability to test for a form of deafness called Connexin 26 deafness, which is the commonest form of genetic deafness, perhaps accounting for 30 to 40 percent of all deaf children, this is a relatively simple molecular diagnostic test. And if it's positive, it can allow you to avoid recommending or doing a lot of other potentially expensive and risky, in some cases, screening tests.
So there is no doubt in my mind that clinical geneticists should get involved after the diagnosis of hearing loss has been confirmed. It doesn't have to be the next day or the next week, but I certainly think that within weeks or months would be appropriate to see a geneticist.

VICKIE THOMSON: And that is -- This is Vickie again, Dr. Nance. That is one of the data items that we have suggested as part of the core data items that states do collect. Not all of the states are aware of all of these data items such as the CDC grants. We're kind of trying to decide what should be (inaudible) and what should be enhanced, and that definitely is something that we would want to try and have states identify.

WALTER NANCE: Yeah.

JANET DEGEORGE: Dr. Nance, this is Janet DeGeorge from Colorado, the parent of a deaf child and also the parent consultant at the Marion Downs Center.

WALTER NANCE: Yes.

JANET DEGEORGE: In terms of having families being referred to geneticists, I think there's a couple of things to look at. Number one is the timing. If it's right during the diagnostic evaluation process, most families are so overwhelmed by the number of appointments and new professionals in their lives. I'm wondering if -- The second thing would be, is the timing imperative in terms of the choices families are going to be making with the information received? For instance, if they're able to find out through a geneticist that the hearing loss of the child may or may not be progressive, that may change the way parents are looking at their choices for communications. So that information is really important in the beginning. If it's information that can wait, you know, three months or something, I think people kind of need to look at the protocol for the timing of when they're referring families. And that would be information families could also -- that would help them maybe -- help them get into a geneticist quicker if they knew that they weren't going to just be getting diagnostic information or information about etiology of the hearing loss that may be information that may help them make choices that are coming up for them.

WALTER NANCE: I think that our group, when we met to consider this, felt that there are so few geneticists in the country that children should not be referred for genetic evaluation of hearing loss until after this hearing loss has been confirmed, you know, until after it's for sure that there's a clinically significant hearing loss. It's -- There are certainly forms of genetic deafness in which establishing a definite diagnosis can have an impact on the management of the child. I'm thinking of things like Usher Syndrome where you're dealing with progressive visual

loss as well as hearing loss. Another issue is the fact that now that we have the ability to test for a form of deafness called Connexin 26 deafness, which is the commonest form of genetic deafness, perhaps accounting for 30 to 40 percent of all deaf children, this is a relatively simple molecular diagnostic test. And if it's positive, it can allow you to avoid recommending or doing a lot of other potentially expensive and risky, in some cases, screening tests.

So I think that -- I definitely feel that the geneticists should get involved after the diagnosis of hearing loss has been confirmed. You know, it doesn't have to be the next day or the next week, but I certainly think that within weeks or months would be appropriate to see a geneticist.

UNIDENTIFIED SPEAKER: Dr. Nance, getting back to something you had mentioned before about some states are thinking of adding some additional genetic tests for hearing loss --

WALTER NANCE: Yes.

UNIDENTIFIED SPEAKER: -- are you looking at coupling those with any of the states that are looking to track hearing through metabolic -- having it reported on the metabolic screening form? I think there are under ten states that are looking to do that now.

WALTER NANCE: They're looking to do what, again? I didn't catch that.

UNIDENTIFIED SPEAKER: They're going to ask for reports from the hospital on the hearing screening results and then record them on the metabolic screening form. And I'm wondering whether there's -- are they looking at some states specifically who are going to want to do the (inaudible) or (inaudible) collecting newborn hearing screening data as well?

WALTER NANCE: I'm not aware of any states knowingly testing for genetic forms of hearing loss in the newborn screen. There is one entity that is screened for, something called biotinidase deficiency, that does result in hearing loss in about 40 percent of cases which can be completely prevented by giving these children biotin tablets, but that is a metabolic disease and was not originally identified as a form of genetic hearing loss.

The group of diseases that I would focus on if I were designing a newborn molecular screening program would be those forms of genetic deafness that are delayed in their onset, perhaps, later in infancy or perhaps in early childhood where you might miss them at a newborn hearing screen. So I view this type of a program as being a compliment to audiologic screening rather than being competitive in any way. The point being that genetic factors undoubtedly play an important role in presbycusis, but I can't see what the rationale would be for doing a newborn screening test to detect people who are going to develop hearing problems when they're 50 or 60 years old. But there would be a very important reason to detect infants who are not deaf at birth but become deaf in the first year of life, for example.

With respect to some recognized forms of genetic deafness, like Connexin deafness, for example, when this condition was first described, it was first found in infants and children and adults who had profound hearing loss. As their brothers and sisters and relatives began to be tested, we realized that the variability and the expressivity was much greater than we thought it was

initially. And I don't think that anybody can put their hand over their heart and say with 100 percent certainty that every case of Connexin deafness, which is the most common form of genetic deafness, is necessarily going to be expressed at birth. It's possible that some cases may not result in hearing loss at birth and would only be detectable if screened for by a molecular test or by audiologic testing later in infancy or childhood.
So that would be a very important piece of information that we would like to know is when this commonest form of genetic deafness is expressed and whether it is ever delayed in its age of onset.

UNIDENTIFIED SPEAKER: One last comment from Minnesota here about the genetics part. As we work to help the families understand the value or the appropriateness of genetic evaluation, just like we've struggled with newborn hearing screening, is reimbursement ends up being a critical part that we often skip over, but yet that, for some families, is a major barrier. Even with the primary care referral, it may depend upon their health plan, in and out of plans, and I'm making the assumption that a genetic evaluation is not cheap.

WALTER NANCE: The clinical evaluation and counseling is probably comparable in cost to the pediatrician's office visit. But you may have diagnostic tests on top of that. I think if these tests are done in a carefully coordinated manner -- for example, if you have a child with so-called non-syndromic deafness, no associated findings -- the first thing to do would be to screen for this commonest form of genetic deafness. That's a relatively inexpensive test perhaps $100-$200, and if that test is positive, it means that you don't have to do lots of complicated tests to see whether they have Usher's Syndrome, to see whether they've got Pendred's Syndrome, or a lot of other disorders.

So I think the tests could be scheduled in a sequencer that would minimize the cost of the evaluation. But, certainly, in our state, insurance companies are now beginning to cover genetic evaluation and counseling to a much greater extent than they did in the past. And I think we have relatively few patients in our genetic clinic where the third-party payers refuse to pay for an indicated referral, evaluation or diagnostic test.

LEE ANN RAMSEY: Thank you all very much for presenting on this call and for joining us today. Are there any last comments or questions?
(NO RESPONSE) If not, our next call will be on November 6th at 2:00 Eastern time. And again, we thank you for joining us and we'll talk to you in November.

[Whereupon, the teleconference was concluded at approximately 2:52 p.m.]