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Larry Thompson
National Human Genome Research Institute
The NIH Undiagnosed Diseases Program: Patient Advocacy Group
May 19, 2008
8:30 am CT

Conference Facilitator: Please stand by your conference is about to begin.

Good morning. Welcome to the Undiagnosed Diseases Program: Patient Advocacy Group Telebriefing hosted by the National Institutes of Health. This telebriefing will last up to 90 minutes. There will be four primary speakers who will provide brief remarks and then patient advocacy group representatives will be able to ask questions.

To ask questions, you can press star and one on your touch tone phone to enter the queue. And you may remove yourself from the queue by pressing the pound key.

This call will be recorded and transcribed.

Now I'll turn the program over to Moderator, Larry Thompson, Chief of Communications at the National Human Genome Research Institute. Please go ahead, sir.

Larry Thompson: More than 100 of you who are expected to join us as the patient advocates who will be joining us on this telebriefing.

Additional material related to this announcement is available on the Web sites of the Office of Rare Diseases. And you can find that by pointing your browser at rarediseases.info.nih.gov/undiagnosed. And you'll find lots of material related to this discussion today. And there'll be information on the Genome Institute's Web site also at genome.gov.

Our expert panel in the order of speaking will be Dr. William Gahl who is the Clinical Director of the NHGRI, of the Genome Institute, and who will be supervising this program. Dr. Steve Groft who is the Director of the NIH Office of Rare Diseases. And Dr. John Gallin who's the Director of the NIH Clinical Center where most of this work will actually take place, which is the NIH's amazing, fabulous, big new hospital.

And we have a special speaker from the patient community. Amanda Young is here from Atlanta to tell us about her experience of being taken care of here at the Clinical Center, and struggling with the disease that initially nobody knew what it was about.

But let me start with Dr. Gahl who'll give us some brief opening remarks and everybody will talk in turn, and then we'll open it up to questions. Dr. Gahl.

William Gahl: Thank you, Larry. Hello, I'm Dr. William Gahl the Clinical Director for the National Human Genome Research Institute of the National Institutes of

Health, the nation's medical research agency based here in Bethesda, Maryland.

I'm pleased to help launch a new trans-NIH program that will serve patients across America who have undiagnosed diseases. They suffer symptoms that the medical profession has been unable to understand and treat. The anxiety of not knowing adds to the turmoil for these patients and frustration for their physicians to try to provide the best possible care. To help these individuals, we are launching the Undiagnosed Diseases Program here at NIH. Their predicament is compelling. And the benefit gained by studying the serious illnesses will add to our fundamental knowledge of how systems of the human body can go awry in ways not yet understood.

This program has two goals: to give hope to the hopeless who are sick, but their doctors don't know what is wrong with them, and to conduct the most fundamental of medical research for the discovery of new diseases. This is after all a research program. Not the straight-forward medical care you would receive in a community hospital.

While the Undiagnosed Diseases Program is a new, unprecedented, Trans-NIH program, it has its origins in NIH's long and proud history of medical research. For many years, patients have come to NIH from every corner of America seeking answers to their scientific and medical questions. Most arrive with some notion of what was wrong with them, usually a precisely defined illness being studied by NIH doctors, not patients without a diagnosis.

For patients whose disease eludes diagnosis, this program offers a chance to have their conditions considered for evaluation here at NIH with a new focus into what their mystery diseases can teach us. For the individuals who

participate, the program will offer expert medical evaluation and potentially helpful guidance for their therapeutic care.

The breadth of basic and clinical research at NIH is comprehensive. The Undiagnosed Diseases Program will offer a new vehicle by which doctors with different areas of expertise can collaborate and optimize our potential for understanding and solving mysterious conditions. The NIH Office of Rare Diseases, the National Human Genome Research Institute and the NIH Clinical Center have enlisted the support and expertise of clinicians in multiple specialties across the 27 institutes and centers of the NIH. The Undiagnosed Diseases Program at NIH offers unique opportunities for discovery within the context of medical investigation into human diseases.

However, we won't be able to help everybody who seeks our care. So for that reason, we've established a fairly stringent referral process to ensure that we have a reasonable chance of helping the people we do bring to Bethesda.

As with all studies at the NIH Clinical Center, the avenue for participation in the Undiagnosed Diseases Program begins with referral from a patient's health-care provider. Patients won't be seen on a walk-in basis. Participants must be referred by a physician or other health-care provider, such as a nurse practitioner or physician's assistant, in their own community into whose care they will return after they are seen at NIH. The referring physician would have to provide a medical summary and medical tests that point to some clue about what might be wrong with the patient.

For example, a patient may have an abnormal laboratory value, a mysterious x-ray finding or a collection of symptoms that usually do not occur together. Clues such as these give the NIH physicians some direction in which to pursue a diagnosis.

Invitation to participate in the pilot will be based ultimately on the medical judgment of a board of medical reviewers here at NIH. They will have the final say about who's accepted into the program. We'll start slow in this initiative. The program is prepared to accept one to two patients per week and as many as 100 patients during the course of a full year - at least initially.

If a patient is selected following physician referral and medical board review, he or she will be invited to visit the NIH Clinical Center and will be offered enrollment in a study for a medical evaluation. The patient will have to provide consent for the investigations. The care will be free for the patients. NIH will pay for travel and lodging. Patients in the program will be evaluated at NIH's hospital – the NIH Clinical Center in Bethesda, Maryland – usually for about a week using the Clinical Center's unique combination of scientific and medical expertise and resources.

Dozens of NIH senior attending physicians will consult on these cases. Their specialties include Rheumatology, Immunology, Oncology, Mental Health, Nephrology, Hematology, Ophthalmology, Neurology, Laboratory Medicine, Pain and Palliative Care, Bone Disorders, Endocrinology, Oncology, Immunology, Dermatology, Primary Immunodeficiency, Dentistry, Genetics, Pathology, Pulmonology, Cardiology, Primary Immunodeficiency, Internal Medicine, Pediatrics and Hepatology.

If a diagnosis is determined, treatment options will be explored, but may not be available. Individuals who are evaluated at NIH as part of this research program will be referred back to their own physician or health-care provider so that follow-up care is assured.

The cases will contribute to a catalog of descriptive conditions. A so-called phenotype atlas for the country. As the experiences of the doctors working with these hardest of cases grow, the team intends to develop a protocol to help other doctors work up a case that is resistant to diagnosis. NIH expects that this program will produce many scientific publications and probably inclusion of new information in textbooks on diagnosis. Finally, some patients will be entered into existing clinical protocols attempting to produce new treatments.

Now to put the Undiagnosed Diseases Program in context to some current related events, I'd like to hand it over to Dr. Groft. Steve?

Steve Groft: Bill, thank you very much. Before I give any comments, I would like to thank first Bill for heading this project with the National Human Genome Research Institute. And a special thanks to Dr. Elias Zerhouni and Dr. John Gallin who have given us this tremendous opportunity to initiate a project. It's a very nice activity that I think will continue to grow into the future.

One of the most frequent requests that I've received over the many years here at the NIH, and in the whole rare disease area has been a request from the patient advocacy groups for the message to initiate a study on their disease here at the Clinical Center. And while this will not initially do this, we can anticipate that perhaps some of these findings may lead to the opening up of new protocols for various rare diseases. So, it is with a great deal of anticipation that we are starting this program. As many of you know, the Office of Rare Diseases coordinates research and information on rare diseases at the NIH, and for the entire rare disease community.

Also, the occasion for this program launch is coincident with a number of other events that are meaningful to the entire rare disease community. This

includes the celebration surrounding the 25th Anniversary of the Orphan Drug Act. It is also the 25th Anniversary of the National Organization for Rare Diseases. Consistent with this is a major conference – the International Conference on Orphan Diseases and Orphan Drugs that will be starting tomorrow. And there's an on-going conference today on the Orphan Drug Act at 25 years – a retrospective and future view hosted by the Food and Drug Administration and the Drug Information Association.

The Genetic and Rare Diseases Information Center funded by the National Human Genome Research Institute and the Office of Rare Diseases report that 6.6 percent of inquiries during the past three years were related to an undiagnosed disease. Also, from a study done in 1988 with the National Commission on Orphan Diseases, approximately 50 percent of patients received a diagnosis in less than one year. It took 31 percent of the patient between one and five years to obtain the diagnosis. And for the other 15 percent, more than five years were required to obtain a diagnosis, including multiple visits to physicians at numerous clinics during that time period.

As I mentioned, all the participants will be seen at the NIH Clinical Center here in Bethesda. I'd like to ask Dr. John Gallin, Director of the NIH Clinical Center to speak to us about this facility, and the role the Clinical Center will play in the Undiagnosed Diseases Program. Dr. Gallin?

John Gallin: Thank you, Steve. And I also thank Dr. Bill Gahl for helping to lead this new exciting program. Better health and health care for everyone depends on clinical research. Medical research is the sole mission of the NIH Clinical Center, guiding all of its activities for more than half a century.

The NIH Clinical Center – the nation's clinical research hospital provides an extraordinary environment for excellence in both patient care and

collaborative clinical investigation. The Clinical Center is the largest hospital in the world totally dedicated to clinical research. Nearly 10,000 new patients come to the clinical center each year from across the country and even abroad. Currently more than 80,000 patients are participating as inpatients and outpatients in the 1,500 clinical research studies being conducted here. And about half of our patients have rare diseases.

Some 1,300 credentialed physicians, dentists and doctor-prepared investigators, along with more than a thousand nurses and allied health professionals work in the Clinical Center. They care for patients, as well as manage and monitor the clinical research studies. About 140 specialized clinical teams made up of dozens of medical specialties see patients at the NIH Clinical Center. This new program will marshal a rich set of skills and expertise already at the Clinical Center to help patients with unusual medical conditions.

Our patients are also truly partners in the process of medical discovery. Patients interested in participating in this research program need to discuss the option with a physician or their health-care provider, such as a nurse or a physician's assistant. Information specialists at the Clinical Center's patient recruitment call center can provide more information about eligibility and what kinds of medical information referring physicians must submit for review by the program's medical team.

The number to call is 1-866-444-8806. You can also reach access through the World Wide Web at <http://rarediseases.info.nih.gov/undiagnosed>.

Along the way towards preparing for the launch of this new program, various individuals came to mind – the young and the old who represent the personal aspect of the clinical research programs at NIH, and now this Undiagnosed

Diseases Program. One such patient, an extraordinary and courageous woman who has joined us today to make this announcement is Amanda Young. Amanda could tell us the story of what difference the diagnosis meant to her in the course of her life, and in dealing with her difficult disease. Amanda could you share your comments?

Amanda Young: I sure will. Thank you, Dr. Gallin. Hi my name is Amanda Young, and I'm 26 years old, and I live in Conyers, Georgia with my parents, Speed and Lisa Young, and my sister, Alex, who are all here with me today.

For most of my life, my parents searched for answers to my medical condition that left me vulnerable to life-threatening infections. No one could tell us why these horrible infections attack my body over and over again. No one knew how to stop them because we couldn't understand why I continued to get them.

My immune system looked as normal as anybody else's under a microscope, except for one small thing: a continuously low white blood cell count.

Even after years of trying, no one could give me a name or a reason of why my life was threatened time and time again. By the time that I was 3 1/2 years old, I'd had spinal meningitis three times, many seizures, and an abdominal abscess the size of a cantaloupe, just to name a few.

When I was eight, they had to amputate my leg. It had started with a small scratch that I'd gotten while I was playing. And overnight the scratch became infected. And as a precaution, I was put into the hospital for a week.

However, two weeks later I was fighting to stay alive in the Intensive Care Unit. I developed gas gangrene and a massive bacterial infection, and I was forced to amputate my leg and hip to attempt to save my life.

After this infection, my parents' search intensified as they became even more desperate and determined to find out what was wrong with me.

My family and I are able to be here today because of the search for answers that had led us to here to the Clinical Center and to Dr. John Gallin. My first visit here was in 1990 when I was only 9 years old. Dr. Gallin made us a promise and he lived up to that promise. He told us that he would never give up on me, and he hasn't.

And on May 13, 2003, we received the magical words that Dr. Gallin and I have been waiting for for 20 long years. My disease finally had a name.

I want anybody who is researching – I'm sorry. I want anyone who is researching for a diagnosis to be able to experience what my family and I did that day. It was so incredible. All we ever wanted was for my disease to have a name and for someone to tell us what was wrong. And Dr. Gallin did that for us.

John Gallin: Okay. Take a breath. You are doing good.

Amanda Young: I have an extremely rare genetic mutation. It's called the IRAK-4 deficiency. My body doesn't create a certain protein it needs to fight bacteria, therefore making me a target for life-threatening infections.

I haven't had a massive infection in several years, which is great. And I have to pay close attention to what my body is telling me all the time. I can't let infections sneak in since my body doesn't automatically recognize the infection. I continue to come to the NIH throughout the year depending on my health, or if Dr. Gallin needs me for more of his studies. Although my disease

has a name now, we don't have a cure or a treatment. So, I will continue to come here for my own studies, and hopefully further medical knowledge to help those at the same time.

It's great to know what they have learned from me could possibly help someone else in this process. That is what life is all about; it's helping out each other. And if what I had suffered through could somehow help someone else not have to suffer, then I am so thankful.

The announcement today of this new Undiagnosed Diseases Program is like handing somebody their life back. Everyone who is sick has hope to get better, and with hope they need help. And Dr. Gallin gave us that help so many years ago. And now today, that hope is being offered to people all over the country. This is the most exciting news that anyone suffering from an unknown disease could hear. Someone is going to try to help you.

Now here at the NIH, they have the expertise, technology needed to help study rare diseases like mine. In an odd sort of way, the NIH is like a home away from home for me. I love Dr. Gallin from the bottom of my heart for all that he has done for my family and I, and continues to do for us.

Thank you all so much for dedicating your time and effort to this new project. You have not only given people a place to come for help, but you've also given us a place to come for hope. And I thank you all so much.

John Gallin: Thank you, Amanda. That was great.

Larry Thompson: All right. So thank you very much to the panel and Amanda, thank you for your wonderful words. And let us – what we'd like to do now is open up the phone to questions.

And so we have, you know, 86 folks on the phone, something like that. I suspect that there are a few of you who have some questions about how this will go, and which direction you'd like to see. What questions do you have about the complexity of this program? This panel will basically answer the questions in turn, and we'll go on from there. So, does anybody out there have a question they would like to start with? Or it was just all so clear that there are no questions whatsoever?

Let's start with Heather Long, has a question from SWAN. Heather?

Heather Long: Yes. Hi, my name is Heather Long, and I'm on the board of SWAN, which stands for Syndromes Without A Name. And my question is – well first I'd like to make a comment.

I wish I had known about the NIH Program when my son was alive. Because all I kept being told was – by the doctors, well we can tell you what he doesn't have, but we can't tell you what he has. And, you know, my son was dying. His disease was rapidly progressive, and none of the doctors pointed me to you all. I asked and I begged – there's got to be a study somewhere, somebody who can help us. And I was just sent home continually, you know, without hope.

And anyway, my question is with you all's protocol for admittance into the research program, have you all thought about looking at cases on a post-mortem review? If there's another trial that could potentially be evolved?

Larry Thompson: Dr. Gahl, would you like to answer that, sir?

William Gahl: Yes, I think that would be under consideration. I think it depends in part on how extensive the medical records are.

Heather Long: I'm pretty – for lack of a better term – anal about my son's medical records. And I have also collected tissue samples, as well, that I have at various labs. Knowing that one day – and I kept them specifically for this reason.

Because after he died, I asked the doctors is there a place where I can send his records, or find some help that would maybe, you know, help other people, and also give us some clues as to what happened with my son? And again, nope, they said there's no place you can send your records.

William Gahl: Well this program accepts medical records from patients who are still with us, and I can accept medical records from those who aren't, especially if there's some possible benefit with respect to the family.

Heather Long: Right. Well and that's, I mean you know, like I said I kept asking while he was still alive and then even after he died. And I was told he was just going to be a file in somebody's box. And, but anyway, I do have extensive medical records. And like I said, I have muscle tissue, skin cells and cerebral spinal fluid.

William Gahl: So why don't you contact us through the number, and we'll go through the procedure.

Heather Long: Okay, well that was my basic question – was if your center would be willing to do a post-mortem review.

I have a 9-year-old daughter, and I know there's a lot of families out there who had one affected child, who might be affect – you know, who has siblings

that might be affected to one degree or another. So that would be extremely helpful.

William Gahl: So this board of consultants will receive this – the medical records and will make a decision in that regard.

Heather Long: Okay. And again, that number was – I’m sorry, I wanted to make sure I have it correct – 866-444-8806?

John Gallin: That’s correct.

Heather Long: Okay. Thank you very much.

Larry Thompson: Anybody else have anything they’d like to add? Okay. I’m going to – so if I butcher anybody’s name in this process, my apologies in advance. But we’d like to hear from Jorge Zamudio from the Alpha-One Foundation.

Jorge Zamudio: Yes. Good morning, everyone. First congratulate all of you because this is a great thing that the community was needing. So, I congratulate all of you.

I just want to – well I have one question. Usually going through the specifics of how this is going to work. So you guys are saying that we’re going to probably have the capacity to received one to two patients per week, probably up to 100 patients per year.

If physicians – you said that would receive referrals from MDs, registered nurses or physician assistants – going through the specifics of how this is going to work. Who can call you to talk to you about the patients? The physician will send the referral in one specific form they have to fill out and

send it to you, and then you will meet and decide whether that patient meets the criteria for involvement?

William Gahl: Yes. That's all correct. I think anybody can call the line though. That telephone number is open to patients and physicians and health-care providers as well. But once the call and the arrangement is made, eventually we need a summary letter from the referring health-care provider and medical records in order for us to review.

Jorge Zamudio: Okay. So most likely in this case, if these patients will be evaluated by different physicians before they can be referred to you guys right? Probably that's what?

William Gahl: Yeah.

Jorge Zamudio: Okay. Good.

John Gallin: You should understand that every record will be reviewed.

Jorge Zamudio: Yeah. Okay.

John Gallin: When we say 100 patients will be seen in the first year, we mean 100 patients we expect in the first to actually come to the hospital to be seen. But some review will be done of each record, and we'll give a response.

Jorge Zamudio: Okay.

John Gallin: So, but we need to have, what Dr. Gahl just referred to, is a good letter or referral that gives us enough information to provide a response.

Jorge Zamudio: Any my last question is – I'm sorry. Is there any way that patient advocacy organizations like ours, like the Alpha-One Foundation, can help you in any how to send this information to many hospitals and institutions? Is there anything that we can to help the project?

John Gallin: Our hope is that this press conference or this conference is a beginning of trying to inform the public to the extent possible that this program exists, so people will know about it.

Maybe Dr. Groft wants to respond.

Steve Groft: Jorge, good morning. I think it's also important for you to publicize this within your own Web site as well.

Jorge Zamudio: Okay.

Steve Groft: And make the appropriate – and this is – really goes for all of the patient advocacy groups is to make links to the appropriate sites. And when I say the sites, it's not just our Undiagnosed Diseases Program, but there's so many other sources of information that people just don't seem to be aware of.

Such as the clinicaltrials.gov Web site that has information about ongoing clinical trials here at the Clinical Center and elsewhere. I think, as was mentioned, by Heather in a previous question, so many people don't seem to understand that there are literally thousands of clinical trials going on worldwide looking for patients, and you can find this by doing the search. And I just encourage all the patient groups to provide those necessary links.

And then there are other sites also with respect to adequate information about so many different rare diseases. The National Library of Medicine and the Institute, and even the patient advocacy groups.

You've all done a tremendous job of developing information, but if it doesn't reach the user and the families and the patients who need that information, you know, we haven't really failed, but we haven't met the goals of what we started out to do.

So, any links and any time you have the opportunity to talk to the members of the media, I think, to give them information about these programs and Web sites will be very, very useful, we think, to patients and families, and the physicians and other health-care providers. I encourage you to do so. I think it'd be very helpful.

William Gahl: I just had one final thing to add, I think, and that is that part of our research program we will see patients who don't have a defined disorder, and may have a variant of a known disorder. And that may require involvement of advocacy groups like yours. For example, if we see patients who might have a variant of Alpha 1-antitrypsin deficiency, and we need comparison or collaboration with some of your investigators and some of your patients. We may ask for your help and that of other advocacy groups to help us to make comparisons and make new discoveries about these variants.

Larry Thompson: So we don't have anymore calls currently. Yes we do. Okay. So, Marsha Lanes from the National Organization of Rare Diseases. Marsha do you have a question?

Stefanie Putkowski: Actually Marsha is sitting with me. My name is Stefanie Putkowski. I'm the nurse with NORD: The National Organization for Rare Disorders.

Larry Thompson: Please go ahead Stefanie.

Stefanie Putkowski: Okay. My question – our question is regarding patients from overseas or from outside of the United States. Just wanted to confirm, are they also eligible for this program? I just wanted to confirm yes or no. Would it be the same class as – for them to have to be considered. In other words, directly contacting – and I mean just like people in the United States?

Larry Thompson: Dr. Gallin can answer that.

John Gallin: So the NIH has a long history of accepting patients from anywhere in the world to participate in studies provided that they meet the criteria of our research protocols.

So this particular clinic that we're opening is a little different in that we're looking for people for whom we don't know the problem. In general, we want this – at least initially, to focus on citizens and residents living in the United States.

However, this does not preclude in a rare circumstance, if a patient has a special problem that meets the criteria that we're looking for for that patient coming.

However, we would not be able to provide transportation for that patient beyond what we call the port of entry to the United States. So, that patient would have to get to the United States, to some city, and then our travel arrangements would occur from there. Does that help you?

Stefanie Putkowski: Yes. That does. I actually – I think we pretty much assumed that. But they would be eligible if they fit criteria? We get a lot of correspondence and phone calls from people outside the country, and that's the reason I'm asking.

Larry Thompson: Sure. Dr. Gahl?

William Gahl: Yes I think the bottom line is they would be eligible. Remember there's a board that's going to review all of these things: the medical records and also ascertain that there's availability of follow-up care if a person were to come here.

But in addition to this program, when the medical records are sent to us, remember that there's about 25 senior consultants who are experts in different disorders. Those senior consultants may have an interest in a particular case on their own outside of this program. So they will have access to those records if a patient applies from abroad.

Stefanie Putkowski: Okay. Thank you.

Larry Thompson: Anybody else? Steve? No, you're good?

So let's go to another question. My screen is refreshing here. Could we go to John Schwartzman, please, from PDSA? John, could you tell us what your organization is?

John Schwartzman: Yes. Platelet Disorders Support Association.

Larry Thompson: Terrific. Your question, sir?

John Schwartzman: Yes, my wife will ask the question. She had the question.

Mrs. Schwartzman: Okay. Will you be providing an announcement either that can be used in our newsletters or Web site, or that will go in the newspaper? Now something that we could bring our to physician and say would you like to help us participate in this?

Larry Thompson: Sure. This is Larry Thompson, so, the Web site, that URL that we gave you earlier will have all kinds of background information on it. Both, so that was again was the

John Gallin: <http://rarediseases.info.nih.gov/undiagnosed>

Larry Thompson: So if you go to undiagnosed, there'll be information there. And also on genome.gov. There'll be all the same information. So you should be able to craft any of the – any kind of messages that you would like to put on your Web site using the background information that we'll provide there.

Just to remind you, my colleague just reminded me that this information that we're giving you today is embargoed until noon today. Because, so we'd appreciate it if you don't put it on your Web site before noon, that would be really great. Because we'll have a media conference call right after that, and we don't want to be putting out the information before everybody has it equally.

But we would welcome you – repurposing all the information on our Web sites and spreading it, and as far and wide as you are able to. So that would be just really terrific.

John Schwartzman: The Web site – the URL be on the NIH Web site?

Larry Thompson: It probably – well, we can talk to our friends at – certainly if you go to the NIH Web site and you type in undiagnosed in the search box on the NIH Web site, you'll get to this page.

Steve Groft: And we'll have it featured on the Office of Rare Diseases Web site. That's where the link primarily will be. So, doing research on the Office of Rare Diseases through the NIH Web site, or through one of the other search engines should get you to us. And then it'll be featured.

John Gallin: It will also be available on the Clinical Center Web site.

John Schwartzman: Okay. Great. Thanks very much.

Larry Thompson: So, by the time we're done with this, you should just type it into Google and you'll be able to find it without any difficulty.

John Schwartzman: Okay. Now we're very grateful that this is starting for many reasons in my family. Not only for myself, but my son who has basically the same disorder and one disorder that they cannot provide a name to.

William Gahl: Why don't you contact me directly? I'm a little interested in that. This is Dr. Gahl. You could probably Google me, too.

Mrs. Schwartzman: We look forward to sharing with you.

Larry Thompson: Terrific. Thank you very much. Okay let's go to our next call. With our next questioner whose name is Dora Maillaro-Tomalonis. And if I may have mispronounced that, please correct me. I apologize.

Dora Maillaro-Tomalonis: No problem.

Larry Thompson: Dora, go ahead with your question.

Dora Maillaro-Tomalonis: My question is if we have somebody who's doing some research on our disorder, and by the way my name is Maillaro-Tomalonis. If we have someone who's actually working on our disorder with us, a researcher at another facility, can we give that information to you?

William Gahl: What information? The information that somebody's working on it? Or the actual data? Or what?

Dora Maillaro-Tomalonis: We have somebody who's working on it. We've been working with a research endocrinologist in California in San Diego, who's been doing some research on lipomatosis disorders Dercum's Disease, Madelung's Disease, familiar multiple lipomatosis and familiar multiple lipomatosis with pain.

John Gallin: What you might do is ask your doctor who's working with you there, tell them about this conference, and if they think it would be helpful to correspond with us, this would be a great source of information to initiate a dialogue and a discussion.

Dora Maillaro-Tomalonis: Okay.

Larry Thompson: So again, the information would be sent into the Medical Review Board and would decide whether there's a natural or.

John Gallin: The – I'd like to just emphasize that the aspect of this clinic that we think is different and is special is that we have this opportunity for over 25 specialists

from different areas to approach a problem. And one of the things we're hoping is that a multi-disciplinary approach to these problems are going to help figure out what these problems are all about.

Dora Maillaro-Tomalonis: Oh I understand. Our disease was named in 1889 by Dr. Francis Dercum, and there's not a lot of information out there, and it's incredibly hard to diagnose. We have one or two diagnosticians that we know very well who understand this disease. And, there's got to be a way to get that information out to other people.

John Gallin: Okay.

Dora Maillaro-Tomalonis: Thank you.

Larry Thompson: Thank you very much.

Dora Maillaro-Tomalonis: You're welcome.

Larry Thompson: So could we go to Amy Clugston, please. Could you tell us what the name of your organization is also and then please proceed with your question? Amy?

Amy Clugston: Yes I'm also from Syndromes Without a Name USA. And I have a question.

Many of the families that come to SWAN, their children see many, many, many doctors. So I wondered in the summary letter and all the records, I would worry that some information might be lacking. And how could we be certain that the right information is being sent to you to review in order to be seen in your clinic?

Larry Thompson: That's a very good question. Dr. Gahl, sir?

William Gahl: Well I think that the summary letter and the medical records are the beginning of a process of almost negotiation in a way. In other words, there's back and forth. We have two nurse practitioners here who are going to be discussing the cases with the referring health-care provider. And as – so, if more information is needed, but there's an interest on the part of the NIH, then more information will be requested.

Amy Clugston: What – sometimes, you know, like if the referring physician is a pediatrician or a geneticist – well if it was a geneticist I would hope that they would have a lot of the information. But if a family's going to their pediatrician and the child sees a lot of medical specialists, I would worry that the pediatrician wouldn't have the right information. So on our part, would it – how could we advocate to the families that – what kind of information that need to tell their pediatricians to send, or how to send that?

William Gahl: Remember the pediatrician's letter - the primary care provider's letter is a summary. And the individual medical records can be obtained by the family individually from all those specialists. That's part of the package. So you have a right to that. And we can help you because there are standard forms for obtaining that medical information to send to us.

Amy Clugston: Okay. Okay.

Steve Groft: Amy, in some respect I think this may even help focus the entire patient and all aspects of the disease. I think for many of the rare disorders what we run into are multiple organ involvement and multiple systems that can be affected by any one particular rare disorder. So, perhaps having that summary prepared by one physician or nurse practitioner or physician's assistant – we're able to bring in the entire picture together a little bit more clearly.

Amy Clugston: Okay.

Larry Thompson: That was Dr. Groft. So all right. Terrific. Let's go to Doug Buckner who's at the – well tell us what the name of your organization is. It's a little truncated on my screen. Doug?

Doug Buckner: Yes this is Doug. I'm with Morgellons Research Foundation, and we would certainly like to thank you all for initiating this. It's been long needed.

My question is what kind of timeline are you projecting for start-up and implementation?

Larry Thompson: Dr. Gahl?

William Gahl: We're accepting phone calls now, and records as soon as the phone call is made.

Doug Buckner: Great.

Larry Thompson: Any other questions? Anybody else wish to respond? Okay. Let's go to Deborah Friedman please.

Deborah Friedman: Hi there. My name is Deborah Friedman and I'm the president of the North American Neuro-ophthalmology Society. And as neuroophthalmologists, pretty much all we do is see patients with rare and undiagnosed disorders. We see very unusual things. And we actually have a Web site and are starting a patient portal for incredibly rare diseases. And I was just wondering if there was somebody there that I could speak to later about the possibility for collaboration?

Larry Thompson: Collaboration around the Web site? Or collaboration.

Deborah Friedman: About patient information who have rare neuroophthalmatic disorders.

Larry Thompson: Steve, you want to answer that? Dr. Groft?

Steve Groft: Yeah, Deborah, thank you. There's several sources here and I think, you know, our Office of Rare Diseases would be happy to initiate the conversations with you. Then I think further conversations with the National Eye Institute and the National Institute of Neurological Disorders and Stroke, in addition to what we're offering in this program; I think that would be in order. And they both have tremendous information departments that can provide you information.

And I think it's also worth while if you haven't done so recently, to come into the NIH Web sites and the various institutes to see if the information that is provided is appropriate and up-to-date. And I think that's a big part of what we'd like to do with the patient advocacy groups. And it is significant because so many people do have ready access to the NIH Web sites, and they look to the NIH as a reliable source of information from all the institutes.

So, I think please send us a note as well from our Web site, and we'll get back in touch with you. Probably not before next week...

Deborah Friedman: Okay.

Steve Groft: ... because of the week that we have going this week.

Deborah Friedman: Is there a neuroophthalmologist on your panel?

William Gahl: Not now.

Deborah Friedman: Okay.

William Gahl: But we have an ophthalmologist and reasonably close ties with the clinical ophthalmology branch of the National Eye Institute. I actually might recommend – can I mention a name? The branch chief for that is Ian MacDonald. So that's one person that you might contact. And you might also contact the Office of the Clinical Director of the National Eye Institute. And that would be Dr. Richard Ferris. Both are really good folks. And I think could put you in touch with the people doing neuroophthalmology.

Deborah Friedman: Great. Thank you.

Larry Thompson: So while the rest of you think about what questions you might want to ask, I'd actually like to ask Amanda a question.

I was wondering if you could describe a little bit for us what it's actually like to come to the Clinical Center here at NIH, and to be cared for and to go through the process of studies and evaluation and those kinds of things. What's that like?

Amanda Young: It's great now. I love to come up here because it's so much – it's more different than anything at a hospital that I've been to in Georgia. Everyone here seems to want to help you. And want to figure out what's wrong with you.

And the bedside manner is completely different than doctors at home at least. And I just thoroughly enjoy coming up here. Even if they put me through a

test that might not be the most pleasant, it – they make it try to be pleasant, and try to work with you in anyway that you can. And I really enjoy – I’ve been here for 18 years now. And I don’t think it’s – it wasn’t the place that I needed to come to, that I would be coming here for as long as I have.

And we’re getting answers that we need that I think other people are going to start coming here for now too, which is unbelievable to me. And I think it’s great.

Larry Thompson: We also have Amanda’s mom here, Lisa Young. What was your experience like in the years, especially when Amanda was so little and you guys didn’t know what was going on?

Lisa Young: That’s the most frustrating part I think with a child who is ill is trying to find someone who can help you. And we were very aggressive in finding different doctors and research facilities who would try to help us. But no one would stay with us. They would do preliminary tests and then they would leave. And after Mandy lost her leg, we told them that we just weren’t going to leave the hospital until they told us somebody that could help us. And that’s when Dr. Gallin came into the picture and we started coming here.

It’s – I can’t say that I’ve always looked forward to coming, because I’ve always known ahead of time what was going to happen, or what she was going to be put through. She looks at it as a vacation each and every time we would come. And look forward to seeing Dr. Gallin, not that we didn’t look forward to seeing him, but it sometimes it was tough.

I mean it’s hard when your child is sick and faces death so many times throughout their childhood. And you don’t know what’s going to happen next. And we saw so many parents around us just give up because they didn’t

know what to do or where to go. And it gets very frustrating and we understand when those people give up. And the thought that now they have somewhere, that hopefully the word will get out and that people will refer them to come here. Their sense of being able to help their child will come back. Because so many parents lose that along the way. And for us, it gave us hope again.

Larry Thompson: That's – thank you very much for sharing. We have – I don't have questions in my queue at the moment. So if there's anybody still on the phone that would like to ask a question, now would be a good time.

And we also do have a fair number of health professionals on the phone. So if there are anybody in the health community – the health provider community who has some questions about how this would work, also now would be a good time.

Amy Clugston is back on the phone with another question. Amy, please go ahead.

Amy Clugston: I have a few more questions.

Larry Thompson: Carry on.

Amy Clugston: And, some of the families that come to SWAN for support have entered into a clinical study for kids with suspected genetic or metabolic disorders. And I just wanted to be able to explain to them what's different about this, or what's the same about this program? If they are thinking about coming to the program.

John Gallin: Well maybe I can start, and Dr. Gahl might want to elaborate.

Larry Thompson: This is Dr. Gallin.

John Gallin: The first is that the Clinical Center is referred to by our nurses as there's no other hospital like it. And by that we mean it's not only a place that focuses on trying to provide patient care at its best. And we think we do a good job at that, but to merge that with the scientific mission of the NIH.

So every patient who comes here, comes here with folks one who have time to dedicate to understanding the disease. More time that is probably available anywhere else. So a doctor can spend an hour or two hours or three hours talking to a family and to a patient because that's so important in understanding a disease. So, while we can't see millions of patients the way a very large city hospital can see, we see patients for many, many hours longer than you'd probably get in other places, and that enables us to understand the problem. And then to apply the most sophisticated kinds of scientific approaches to questions to try to understand what the problem really is and to try to identify new lessons learned that can not only help the patient who comes, but others, sometimes others who have common diseases that relate to the problem the people with rare diseases have.

So this is a place that's very different. It's a place where sometimes patients will stay for a long time. We have a full school at this hospital, from kindergarten through high school, because one of our goals is to make sure that children don't fall behind in their education. We have recreational therapy facilities that you don't see generally anywhere else. It's, you know, sadly for some children it becomes a home away from home, but that's also a very good thing because it provides hope.

Dr. Gahl would like to elaborate.

William Gahl: Yeah. Just one other thing is that sometimes the existing protocols like the one you mentioned perhaps are run by an individual principle investigator who has an individual area of expertise. And this new program allows a multi-disciplinary approach. It allows some intellectual synergy on the part of the consulting physicians, so that's a breadth of perspectives could be offered and perhaps the entire constellation of symptomology could be addressed.

Amy Clugston: Okay. That explains it well. One more question as well. I guess when I'm talking to the families, I would like to know an idea of how long the review process will take. I can imagine that when I talk to the families they're going to – you might have a lot of families coming to you, and quickly probably. So, how long do you think that review process will take?

William Gahl: Our plan is to be able to get back with the review in six weeks after all of the data are in. Now of course sometimes things are pending, and sometimes patients think that their physicians have sent the data in. So there's a little bit of uncertainty there. But once we have a complete record here, and bring before our board, we expect to get back to people within six weeks.

John Gallin: But we will get back to people very quickly to tell them the status of the information that we've received. So, there will be a post card or notification as soon as the materials first arrive saying that it's arrived, and then if there are problems in terms of getting more information, we will have correspondence.

Larry Thompson: Dr. Groft?

Steve Groft: Amy, you bring up a really significant issue. And in that being the knowledge and leadership of the patient advocacy groups. We sometimes really fail to acknowledge all the tasks and responsibilities that the leaders are asked to

perform. And so, for the individual, you know, the President, Executive Directors or the Director of the Scientific or Medical Advisory Board, I think it's so important to stay in touch with the research community, with our program here, and various specialists, so that you understand the protocols. And there are many people who are willing to take the time to explain the protocols and to explain this program to you in greater detail, so that you're very clear.

And I think that's a preface of what we're trying to do today is to initiate these conversations and discussions, so that you're very comfortable talking to the patients and the families about any protocol, any study that a patient or a member of a family might consider joining. And so, please take advantage of the resources that are available and just keep asking questions. And I think you'll become very clear as to what is going on and what the future looks like as far as the clinical studies.

Because for many of the rare disorders, we, you know, we don't have treatments for many of these diseases, but I think what to mention is that we can provide hope. That perhaps by coming and participating in studies, that we do develop the information that can be more useful as we move forward into the future.

Amy Clugston: Yeah. Thank you very much.

Larry Thompson: Okay. So let's – so we have a few questions in the queue now. So, could we go to Marsha Lanes at NORD? Marsha?

Jackie Ross Planes: Jackie Ross Planes also with NORD. First of all I'd like to thank you very much.

I see first-hand with the phone calls I receive everyday the constant need. There is – for what you’re setting up here in this new venture that you’re about to embark upon. I thank you so much.

And I just wanted to clarify with you – we receive phone calls constantly from people who are struggling to try to receive a diagnosis. And I certainly want to make sure that we use discretion in people that we send to you for your resource. I was wondering if there were any specific criteria that stand out that we should look for before needlessly sending people over to you. Because you can end up being inundated from what I have seen with the – how many people are out there that have had so many problems getting a diagnosis for a rare disease.

Larry Thompson: Dr. Gahl?

William Gahl: Well I think for one thing we’re going to be inundated anyway. But, on the other hand, there are a few written criteria here, and, you know, and one is that the patient is able to travel to us. And the other is that they have a health-care provider who can give us the information that we need in order to make an evaluation. And there isn’t too much else. I suppose that you can use some of your own discretion for some of the intangibles as well.

John Gallin: I’d just like to add one thing. One of the things we really look forward to is establishing a partnership with the patient’s primary physician. And it’s important that some form of primary care exists for the patients. So if a patient has no doctor and has no team, either we have to identify a team that’s going to work with us – and people like you can help do that. That’s absolutely critical.

Steve Groft: And also I think many times what the patients are confronted with are the travel barriers that some of them face. So the Clinical Center does have different policies toward travel to the protocols I think is important. Again, to establish contact and to see what's going on. And there are other services such as Angel Flight with Mercy Medical Airlift and others that can also help for shorter trips. So, I think don't let the travel issue restrict gaining access to this protocol, or to any others, because there are services available that can help facilitate this aspect of just getting to the clinic or treatment centers.

John Gallin: The same is true for lodging. We have a children's inn on the campus. We also have a family lodge on the campus which can house patients, and the program can cover those costs. And then there are lots of hotels, and this particular protocol that we're describing can accommodate the costs for housing for a patient and for one guardian or an adult to accompany a small child.

William Gahl: If it's medically needed.

John Gallin: Right.

Larry Thompson: All right. Let me just remind, you know, everybody who's on the phone, that if you do want to ask a question and you came on late and didn't hear the instructions earlier on, you push the asterisk 1 key to get in the queue to ask a question.

Let's go to Leslie Hanrahan of the Lupus Foundation.

Julie Veners: Hi, this is actually Julie Veners at the Lupus Foundation. I just wanted to, on a personal note, first thank you for all that you've done. I have a daughter with a mitochondrial disease, and I know Steve Groft very well. We go back many

years. My daughter, Brielle, is now almost 14, Steve, if you can believe that. And 14 years ago, it was unbelievable where we were with mitochondrial disease, and where we've come. And that's in great part to Steve's work over at the Office of Rare Disorders.

So I really need to talk to you about lupus though, because lupus as you know is not necessarily a rare disease. Many people are diagnosed properly and many are undiagnosed. And it requires a multi-disciplinary approach, and as we know, it's a disease of unknown origin. And as you probably also know, we haven't had a new drug that's FDA-approved in over 40 years, so it's really difficult for our patients to get the proper treatment.

And I'm wondering if one of you might be able to address how lupus fits into what – into you're new work?

John Gallin: We have at the NIH a program studying lupus through the National Institute of Arthritis, Musculoskeletal and Skin Disorders, and the Clinical Director for that Institute is named Dan Kastner. And you can certainly correspond directly with Dr. Kastner through the Web site of the – called NIAMS – for questions about lupus.

There may be some variants of lupus that would represent something new because lupus is really a spectrum of disorders. And so if they – a referring physician thought that a patient had some unusual variant of lupus that warranted some evaluation here, that's the kind of problem that I think should be referred to this new clinic.

Larry Thompson: Okay. Thank you very much. So could we go to Laura Meints, please? And could you tell us what your organization is? I just have Office of Research on my screen here.

Laura Meints: Absolutely. I'm Laura Meints, and I'm with the Office of Research and Women's Health at the NIH. Thank you for holding this teleconference. It's been a good thing.

My question is how – you mention that 10,000 patients per year who come the NIH's Clinical Research Center, about half of them have rare diseases which presumably are going to involve either consultations with other disciplines, or some other protocol along those lines. How is your new program different from the existing system? And I didn't hear gynecology mentioned as one of the specialties. I was wondering where women's health fit in?

John Gallin: That's a very good question. And I think the major difference here is that most of the – all of the other programs that are already in existence at NIH are highly focused program activities within a single institute with investigators and clinicians who address the problem from their personal interest and talent. What's different in this clinic is that it's a multi-disciplinary clinic involving – we believe the world's best people from 25 different institutions attacking a problem. And so, these problems that come to us will be viewed from a multi-disciplinary perspective, which we hope will add a real intellectual power to the effort.

And certainly, all women's issues and all men's issue for that matter, and all children's issues and all issues of the elderly will be considered by this clinic.

Laura Meints: So I just want to clarify.

That - then say a primary provider with multiple consultations, and presumably those people are talking to one another, you mentioned the richness of the dialogue that's one of the reasons why people should want to

come to the NIH for their care. This would be a more formalized system where you would have specific meetings, or all the providers would be there at least once? Or, how is the implementation going to work?

Larry Thompson: Dr. Gahl.

William Gahl: Well as we receive the medical records and the referral letter, this will be sort of encapsulated and then presented before this group of about 25 consultants on a monthly basis. And from that consultation meeting will emanate decisions on whether to accept the individual patients.

Laura Meints: And then once they get there, how is this multi-disciplinary clinic going to function? And how is that different from having a primary provider who has responsibility over the patient, and multiple consults in different areas? They're presumably talking to one another as well.

William Gahl: There will be a primary investigator taking charge who will be in close contact with other consultants on this. And that can include the gynecological consultants who will be available here at this Clinical Center.

Larry Thompson: Great. Thank you very much. So let's move on to another question now. We have a bunch in the queue.

So, could we go to Wendy Chaite please? With the Lymphatic Research Foundation.

Wendy Chaite: Hi. Thank you for having this call, and I applaud these efforts. I understand that the criteria to apply will be posted on the Web site, but is it public, what the facets in criteria for selection will be? That the consulting board and reviewing, because I'm assuming there's going to be very diverse

applications, are they looking at the NIH portfolio? Are they looking at the length of time of undiagnosis? Or the complexity of the disease? The extensive local support? You know, the window that this rare disease provides to common disease? Is there a way of planning out what those facets are?

William Gahl: They're very individualized. And but the bottom line is if there's enough evidence in an individual medical record where we think that we can be of some benefit, and we think that we can learn things about this, those are the bottom lines on this. There's a lot of medical judgment involved by 25 different specialists. So, no, I don't think there are great criteria that we can tell you about. They're very diverse just as you mentioned.

Wendy Chaite: And when it's an issue of like sort of the tools and the technologies to evaluate. Like say for example, Lymphatic diseases – if there's not currently lymphatic imaging available, would that preclude you from, you know, considering it? Because you may not have the tools to evaluate and diagnose?

William Gahl: No, I don't think so. I think that we will address undiagnosed diseases at different stages at which those individual disorders are at this time.

Larry Thompson: Dr. Groft?

Steve Groft: Wendy, good morning. Good to hear from you. It's important, too, you know, that we are going to be making available these FAQs to you and to the public that you can look at, and I think that will help guide you.

But as always, the important aspect of this is the personal communication that's going to be required from you and from the patient and the family to the program. Just to talk about how this will be implemented, I think we can't

overestimate the value of this. Like so many other things, it's the communication that frequently fails. And so I think we're encouraging people just to continue to follow up on a regular basis and at the timelines that have been suggested. And so that answers can be provided back to the families and to the individual patients.

Larry Thompson: Great. Thank you very much. Let's move on to the next question – questioner. Roulette Smith. I'm not really sure I'm pronouncing your name correctly.

Roulette Smith: Roulette.

Larry Thompson: Roulette. Okay. And your organization?

Roulette Smith: Institute for Postgraduate Interdisciplinary Studies. It's your basic research firm.

Larry Thompson: Great.

Roulette Smith: My question is very simple. I've been in on pioneering research in computer science in a number of areas since the early 60's, and one of the things that makes it really work is the way that the information is disseminated once you make your discoveries.

So the question is, is how will this program now present its information? Will it be only through the main stream journals, you know, like the New England Journal of Medicine or the Lancet? Or will there be technical reports to keep people informed of what's happening?

For example, in listening to (Amy's) case, I was fascinated and was – can see where it could have some implications for some of the things that I'm doing. And yet, it probably hasn't really reached the critical stage where it's been published yet.

John Gallin: So, that's a terrific question. And the whole issue about how do you get scientific discoveries accessible to the population is a question that the NIH as a whole has been struggling with, as has the Congress, believe it or not.

And I think the way we would handle your question is that only information that has been assimilated into a concrete enough state so that people believe it's correct information, would be shared. But once the information is ready for sharing, it will be put in a publication.

But the new twist that has just happened in the last couple of months is that the data will be accessible not only in the journal in which it's published, but in a public release within a certain time frame after that journal comes out through something called PLOS, which is going to be a public Web site where all publications, no matter what journal they are, are posted and which anybody will have access to at no cost. So I think that's going to help with the dissemination of information.

But we would not disseminate information if it wasn't in a format that was – we were confident was real information.

Larry Thompson: Great. Thank you. Dr. Groft.

Steve Groft: And again, it's very important for the individual patient to check if it is the name of a disease in which there is an organized support group, and then for us here at NIH to communicate this information back to the individual,

especially organizations that might run into the symptoms of the individual diseases.

So, there are many aspects of this that will be developed and information that will be disseminated widely just to inform the medical and medical community primarily, but also, for the patient groups. We find many of the individual patient groups are the ones who can identify a select group of patients with special symptoms that the entire, you know, that most of the members of that organization they don't exhibit. But they can always pick out some patients that are "a little different or exhibit a little different symptomology than what others haven't." And this is where I think some of the genetic variation we may start to recognize and identify this even more as we go along.

So, the value of this information will just continue to grow. It's not just a one shot event, but I think just to be able to follow that patient over a period of time also would be very very helpful.

John Gallin: There's one more piece of information that relates to your question. And that is starting in the fall, the site clinicaltrials.gov which lists all clinical studies that the NIH sponsors, will not only be reporting what trials are under way, but when studies finish their story and get closure, they will be required to post on this Web site the findings. So even negative findings that might not get published in a major journal will be posted for the public to view. And that's going to be a major, I think, improvement in sharing of information.

Larry Thompson: Great. So let's go to the next question. Marianne Genetti from In Need of Diagnosis.

Marianne Genetti: Hi, yes. I can't tell you. I can almost cry. The hope that you're going to offer people.

And the one thing that not many people have mentioned is the hope it offers to physicians. We have people who call with desperate stories and they said 'And I won't tell the doctor everything because he'll let me go and not see me again.' We have one person whose out in California has seen top specialists at UCLA. And they keep saying, don't come back it's not my specialty. So, this has not – this has enabled physicians to work with patients because now they have recourse.

I want to follow up on the woman who mentioned mitochondrial disorders. Which department would mitochondrial disorders and fatty oxidation disorders – under which category would those disorders fall at NIH in your existing clinics?

Larry Thompson: Dr. Gahl.

William Gahl: Those are classic metabolic disorders.

And now if it turns out that because the symptomology covers different organ systems. Mitochondrial disorders are largely neurological for example. And say metabolic people like endocrinologists or NIDDK might handle some of the fatty dehydrogenase deficiencies, which is basically what you're talking about, but these are metabolic diseases handled by biochemical geneticists.

Marianne Genetti: And is there a certain department that biochemical geneticists are found in?

William Gahl: Yeah, mine. It's the National Human Genome Research Institute. And we have several board-certified biochemical geneticists.

Marianne Genetti: Wonderful. Wonderful. Thank you very much for taking my call. Thank you for being there.

Larry Thompson: Let's go to the next questioner, Jennifer Mandell. Jennifer, and would you tell us what your organization is please?

Jennifer Mandell: Hi. The organization is called About Madelung's. And the disease that it works with is primarily Madelung's Disease. However, there was a person who spoke earlier, Dora Maillaro-Tomalonis who is with Dercum's Hope, and Dercum's Disease and Madelung's Disease are sort of related through the fact that they're forms of lipomatosis, and we have the same research physician working with us out in California for both groups.

My question is about how the patient advocate can – with a disease that I have, it's got a lot of symptoms that don't always seem related to physicians. So when patients contact me, I often end up like getting stuck in this role that's kind of like a Managed Care Coordinator for them.

And I think what happens is that the physicians, if they are willing to even contact you guys on behalf of the patient, the problem's going to be that the physician isn't going to have all the information necessarily. And sometimes the patients themselves don't realize what all is connected. For example, with Dercum's Disease, almost everybody has some degree of night blindness. But it often gets totally overlooked. And then there are diseases that only show up in a handful of people along with Dercum's or Madelung's such as ankle spondylitis, or a clotting disorder or scleroderma.

We don't have a lot of people who have that kind of relationship with a physician or health-care provider – primary health-care provider – that I can

envision either being able to or being willing to get all the information together and get it to your group. And I think that's going to be the biggest barrier I'm going to face.

John Gallin: So I think, you know, you raise a great point. And one of the things that we think we do very well here is a phenomenon called phenotyping, and that's describing in great detail both the clinical components of a disease, as well as the molecular and genetic and radiologic components. So the way you start is you – the patient gives you the history. And then go back and forth and you try to get as rich a history as possible, and look for associations.

So for example, some of the symptoms you just described, you wouldn't normally find perhaps in one person several of them. But our hope is that we have people here who are thinking in those terms who will see connections, and then ask for additional information to try to pull it together.

So I'll let Dr. Gahl elaborate as well.

William Gahl: Well I think your issue is how do they get into this program in the first place.

Jennifer Mandell: Yeah.

William Gahl: Yeah. So, there is a large wholeness of responsibility on the part of the family and the patient in this. They're going to eventually – and your organization can help with that a lot I think in marshalling the forces against this challenge, which is to say get the family to be able to obtain the medical records from the specialists besides the primary care person, to put together this package.

So that's something that we can help with a little bit, but with all the patients who are going to be able to produce complete packages, we may be

addressing those things, you know, as well. So, I think you do have sort of a job before you, and so does the family to do this.

So let me mention one other point. This program and entry into this program is only one avenue of support for people who have disorders like your groups. The other is to try to let's say solicit the collaboration of a possible expert in the field or let's say potential expert. Some, perhaps, young investigator even here at the NIH who might be interested in seeing 10, 20, 30 individuals who have the same type of disorder, and to define that disorder with all its variations as you mentioned

Larry Thompson: Great. Thank you. Thank you very much. So we have two more questions we'll go to. So we won't take anymore questions and when we're done with these three we'll bring this to a conclusion.

Can we go to Michelle Manion please from the PCD Foundation. Can you tell us what PCD is, please?

Michelle Manion: Yes. It's Primary Ciliary Dyskinesia.

Larry Thompson: Thank you.

Michelle Manion: First of all, thank you very much for this program. It's wonderful. As the parent of a child that waited for a diagnosis for seven years, this is just exactly what's needed.

My question is – I have two of them and they go to accessing resources.

If you are part of a group that's already fortunate enough to be involved in a focused NIH study, but people that have the diagnosis and have been enrolled

in this study and are then undiagnosed. Would these be appropriate people to forward into the other – into your program?

Larry Thompson: Dr. Gahl.

William Gahl: I don't quite understand you. You said that they have the diagnosis and then they're.

Michelle Manion: They've been given the diagnosis and then when they participate in a more rigorous research study, there's a lot of misdiagnosis of our disorder.

William Gahl: Oh sure. So then the diagnosis is taken away from them.

Michelle Manion: Right. And they're clearly sick.

William Gahl: Yeah.

Michelle Manion: But nobody can figure out what it is.

William Gahl: The answer is yes.

Michelle Manyon: Okay. And so the researchers that are involved in other focused programs will know about this program and will be able to access these resources?

William Gahl: Not necessarily. I think, you know, that's our job. And it's the advocacy group's job to really advertise that.

Michelle Manion: Okay. And the second question has to do with – and I'm just kind of clarifying. It sounded to me like you've been saying that people with a known diagnosis, but with unusual features are also people that you'd want to see

through this program? Or would it be more appropriate for them to stay with the more – the focused research?

William Gahl: So, this is a subjective issue. So that we would be willing to receive some of those records.

Michelle Manion: Okay.

William Gahl: And the panel will sort of try to decide that. And many times the result might be a suggestion that there's a focused researcher who can address this better.

Michelle Manion: Okay. Thanks.

Larry Thompson: Terrific. Thank you very much. We'll go back to Dora Maillaro-Tomalonis. I'm still doing that badly. I'm sorry, Dora. Please go ahead.

Dora Maillaro-Tomalonis: No problem. I'm with Dercum's Hope. And the question that I had is – I've heard a lot of medical people involved in all of this. And I was wondering if along the way if you found any complementary or alternative medications – medicine programs that would help the patients for palliative care? Will you be using those resources?

John Gallin: The answer to that question is absolutely yes. We have a pain and palliative of care program at the Clinical Center. We also do – we offer traditional medicine such as acupuncture, tai chi even labyrinths here, and we integrate that with the standard of care. And we actually study those approaches to try and get objective data in terms of their efficacy. So the answer to your question is yes.

Dora Maillaro-Tomalonis: Okay. Thank you.

Larry Thompson: Terrific. Thank you very much. And for our last question, Doug Buckner, please, from the Morgellons Foundation – or Research I mean.

Doug Buckner: Yes. Thank you. At the present time the CDC has initiated a study partnering with Kaiser in California to look into Morgellons Disease. Does it have any impact or effect on patients applying for your program?

Larry Thompson: Dr. Groft.

Steve Groft: Yes. Doug, thank you. I think that's an important study that they've initiated there in California. So, you know, we always like to find the results of the studies so we can use those in a more appropriate fashion. So, I'm not sure exactly what we'll be able to do, but I think we will be in touch with the folks who are conducting the study, the CDC and then the local institution with the Kaiser Hospital Organizations that are involved in this. I think before we do too much we want to establish contact with them and see how they're progressing, and what information they might be able to share as well.

Doug Buckner: Okay.

Larry Thompson: Great. Okay. That brings to a conclusion this Patient Advocacy Call.

I'd like to thank everybody who participated in this. It was very excellent discussion and most interesting. I'd like to thank my panel members who have participated in this, and have given such good description.

And we'll bring this to a conclusion. Thank you very much.

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