SOCIAL SECURITY ADMINISTRATION

COMPASSIONATE ALLOWANCES OUTREACH HEARING
ON RARE DISEASES

Washington, D.C.
Tuesday, December 4, 2007

The Outreach Hearing on Rare Disease began at 9:21 a.m.

BEFORE MEMBERS:

MICHAEL J. ASTRUE, Commissioner of Social Security
STEPHEN GROFT, Director, NIH Office of Rare Diseases
FRANK CRISTAUDO, Chief Administrative Law Judge
DAVID RUST, Acting Deputy Commissioner for
Disability and Income Security Programs

Reported by: Cindy L. Sebo, RMR, CSR, CRR, RPR

C O N T E N T S

OVERVIEW OF RARE DISEASES

RARE DISEASES AND THE ADJUDICATORY PROCESS

MS. BRAUNSTEIN: Good morning. I'm Diane Braunstein, and I'm the director of Compassionate Allowances and Medical Improvement for the Social Security Administration.

And before we begin the hearing, I just have a few housekeeping items I wanted to go over.

In the event that there is an emergency sounding of an alarm, people should exit through the back door over there (indicating), outside, proceed to the end of the block, turn right and go under the underpass and wait for instructions.

Second of all, no cell phones can be used in the main hearing room. No food is allowed.

For people who will be testifying or speaking, the sound system is not working optimally, so please speak rather loudly.

And the last thing is that we are here as the guests of the International Trade Commission, represented by Tawana Braxton over

here, and we appreciate their hospitality.

MS. BRAXTON: Thank you.

COMMISSIONER ASTRUE: Good morning.

My name is Michael Astrue, and I'm the Commissioner of Social Security.

Welcome to this outreach hearing on compassionate allowances for people with rare diseases.

I appreciate your joining us at this critical time as we try to improve the way we make decisions about disability claims.

Our top priority is to reduce the backlog of disability cases. This is America, and it is simply not acceptable for Americans to wait years for a final decision on a claim.

We are overdue for change, and we are committed to developing processes that are as fair and as speedy as possible.

To achieve this goal, we have taken, or will take soon, a number of steps to better manage our workloads. The program includes four components: one, accelerating reviews of cases

likely or certain to be approved; two, improving our hearing procedures; three, increasing adjudicatory capacity; and, four, increasing efficiency through automation and improved business processes.

Today's hearing focuses on the first of these components, accelerating the review of cases likely or certain to be approved.

When Congress first enacted Title II, to a large extent, it viewed the program as a fairly limited one, directed primarily at a fairly small number of middle age workers who needed a bridge to early retirement.

Today, our disability programs are far larger and more diverse than Congress initially envisioned.

Title II and Title XVI are critical to people from all walks of life, including children born with serious defects, wounded warriors returning with traumatic physical and psychological injuries, and the homeless and many others.

By comparison, the way that we process medical information to define disability has not changed in any fundamental way for decades.

We divide our listings into 14 body systems, and we only update those --

COMMISSIONER ASTRUE: I'll try to speak a little bit more close into the microphone. I was forewarned that there had been some microphone issues.

Can you hear me in the back, anyway?

UNIDENTIFIABLE VOICE: Yes.

COMMISSIONER ASTRUE: Yes.

Okay. Good.

All right. We don't usually overhaul the listings unless we do comprehensive overhauls, which sometimes wait decades.

Our recent release of updated digestive regulations, which include diseases of the liver and colon, were last revised in 1985.

We've also tended to focus on large diseases and conditions. This model worked perfectly fine in 1970, but it doesn't work today, and it surely will not work in the coming decade as we process a predicted 26 million disability claims. We need to be more efficient and more accurate.

When people don't receive their benefits in as timely a manner as possible, it undercuts the contributions they have made to the system and can continue to make to our society.

Getting people benefits quickly is the right thing to do, especially since claimants are often in medical and financial distress.

Recognizing distress and wanting to alleviate it is how the dictionary --

UNIDENTIFIABLE VOICE: Excuse me. Is someone on line?

MS. WILSON: Yes.

UNIDENTIFIABLE VOICE: Ma'am, it is very poor quality. I do apologize, but we're getting about every fourth or fifth word that he

is saying.

MS. WILSON: Okay. We might be having a technical difficulty at this time.

COMMISSIONER ASTRUE: Could -- could you tell her that every time she calls in, she's interrupting the hearing?

MS. WILSON: If you wouldn't mind, every time you call in, it's actually interrupting, so -- there are some transcriptionists taking notes that we will be posting on line. If it's -- there's difficulty with actually hearing the conference, it will be on line, available later after the conference.

UNIDENTIFIABLE VOICE: Okay. Now, you do realize that it is being recorded on our end. So --

MS. WILSON: Yes.

UNIDENTIFIABLE VOICE: -- it's just -- okay, all right. Okay. We will let them know.

MS. WILSON: Thank you.

COMMISSIONER ASTRUE: I -- I was talking to the staff right before we started, and

I said, well, you know, when we look back on this after two days, I'm sure there will be a few kinks. I just didn't think they'd happen in the first two minutes. So I apologize to everybody.

(Laughter.)

COMMISSIONER ASTRUE: Many of you know from personal experience -- I know I do -- that uncertainty during the long wait for decision on a family member's case can make a bad decision -- a bad situation even worse.

We're trying to change our business model based on some very simple propositions. First, we need to update our listings much more often, and we need to greatly expand the number of diseases and conditions which are included in our listings.

Guidance for one rare disease affecting 50,000 Americans will not make a significant change on the overall process. But if, over time, we can do it for between 100 to 500 rare diseases, we start to make a real difference.

These are precisely the types of cases

where examiners are uncertain as to what to do. This uncertainty leads to requests for time-consuming and expensive consultations often performed by doctors that are unfamiliar with our rules and regulations.

In particular, this is why our first hearing focuses on rare diseases. If the process can be approved for children and adults with rare diseases, it can be approved for everyone.

Second, we need to fast track the cases that are certain or near certain to be allowed, and with our new electronic systems, we can automatically and with precision pull those cases out of the queue for fast processing.

Our initial test of this concept in New England was highly successful, even though the model only included a handful of cancers, ALS and low birth weight babies.

If we do the hard work of identifying the hundreds, perhaps thousands of similarly situated diseases and conditions, which is what you're all here today to help us with, and for

which I thank you, then we may be able to process up to 15 percent of our cases in days or weeks, instead of months or years.

Our quick disability determination system, which is one method we are using to fast track cases, has helped us decide cases in the New England region in an average of 11 days.

That system has just gone national, and we're going to drive the current 3 percent threshold for likely allowances up, with your help, in 2008.

QDD deals with cases that are extremely likely, more than 90 percent, to be allowed.

We're also moving to create a category of -- of conclusively presumptive disabilities called "compassionate allowances"; in other words, diseases and conditions where we know, by definition, that the patient will be unable to work for at least 12 months.

Third, we need to tap into medical advances of the past decade to make some of our

current high volume cases automatic and easy.

There have been stunning developments with imaging and biomarkers that should help us correlate objective information with the functional capacity determination necessary to make decisions regarding disability.

Can we measure progression of MS by measuring neuronal scarring with MRI? Can the newest imaging machines help us tie blood flow to the heart with functional capacity? We have never investigated the answers to these types of questions, but we have started.

As you listen to today's witnesses, as you hear testimony from medical experts, you will hear testimony from medical experts from NIH.

Over the past several months, SSA has been working with the National Institutes of Health to lay the foundation for a partnership designed to provide our adjudicators with modern, up-to-date tools and the cutting edge medical information needed to expedite disability claims. Dave Rust, Glenn Sklar and Diane Braunstein at ODP

and many other SSA and DDS staff across the Nation have been working hard to move our disability initiatives from concepts to realities by the middle of next year.

As terrific as their work has been, we cannot do it alone. Already, we've received tremendous support from our partners in the state DDSs, advocacy groups, NIH, OMB and many others. We will continue to need this help as we move forward.

A few minutes ago, I mentioned a partnership with the National Institutes of Health.

As part of that effort, joining me on the panel to my left is Stephen Groft, director of the NIH Office of Rare Diseases. Steve is taking a great deal of time out of his busy schedule to attend the hearing. I'd like to afford him the opportunity to make a few opening remarks.

Steve.

DR. GROFT: Thank you very much, Commissioner Astrue, for this opportunity to join

you at these hearings on compassionate allowances for patients with rare diseases.

You know, back in 1987, we had what was called the National Commission On Orphan Diseases, and our very first meeting, as we were traveling into Washington to convene the meeting, the snowstorm had started that day, shut down Washington maybe for two or three days, and we had a captured audience. So I think we will release the audience here after two days.

But that was probably our best meeting that we had as a -- as a commission. We were able to bring the thoughts together. So sometimes through adversity, some very, very good things do occur.

More importantly, I would like to express a sincere thank you for the willingness to address this concern.

This need for special consideration for rare diseases was expressed by the National Commission On Orphan Disease at the Department of Health and Human Services, convened

at the request of Congress, and submitted a report back to Congress in 1989.

A review and discussion of rare diseases is extremely difficult. There are approximately 7,000 inherited and acquired rare diseases affecting between 25 million to 30 million people in the United States alone.

There is no one predictable pattern of progression of many of these disorders that may eventually lead to disability. However, for many other rare diseases, the progression can be predictable, and it is extremely dramatic.

There are many distinguished presenters who have indicated a willingness to provide their experiences in the form of testimony during the next two days. I'm sure you've heard from many others who are unable to join us today.

When I use the term "distinguished," it refers to the commitment and accomplishments of these individuals -- these individuals have made to support research and information development and dissemination activities to advance the

knowledge about their rare disease.

The patient advocacy groups are now considered to be partners with the research community in their quest for diagnostics and treatment interventions.

We will hear numerous suggestions for possible paths to follow as you determine the best choice to address the issue.

I think I can say with a certain confidence that whatever paths you choose, there will be numerous clinicians, researcher -- researchers, experienced lawyers and patient advocacy groups supporting your position and assisting in ways beyond your imagination.

They have done so for research during the past 25 years since the Orphan Drug Act was signed in 1983, and I would anticipate the same type of -- of commitment and cooperation in -- in this endeavor as you move forward.

I would also like to compliment the Social Security Administration staff members who have worked diligently to address this -- the

issues surrounding the proposed program. They have generated the excitement that is necessary to sustain this initiative.

We, at NIH, including the institutes and -- and the categorical research institutes and centers and the National Library of Medicine, look forward to the opportunity to continue the collaborative efforts to help meet the needs of patients with rare diseases and their families, who frequently are the primary caregivers for so many with rare diseases.

I -- again, I thank you for this opportunity to participate and I -- I -- I -- I don't know of any other initiative in the past 10 or 15 years that has generated this much excitement in the rare disease community.

So you -- you have your willing partners, and -- and we're looking forward to the outcome of this meeting and other actions of the Administration.

So thank you very much.

COMMISSIONER ASTRUE: Thank you,

Steve.

Flanking me on the right and left are Dave Rust and Frank Cristaudo.

I'd like to give them just a -- a chance to briefly introduce themselves and tell them what they do -- tell you what they do.

JUDGE CRISTAUDO: Thank you, Commissioner. I'm Frank Cristaudo, Chief Administrative Law Judge for Social Security. I've been an administrative law judge for 17 years as a judge and a hearing office, as a hearing office chief judge, as a regional chief judge, and now as the chief judge under the executive leadership of the -- of Lisa DeSoto, the Deputy Commissioner for the Office of Disability Adjudication and Review, responsible for management oversight of the -- of the hearing operation for Social Security.

Thank you.

ACTING DEPUTY COMMISSIONER RUST: I'm Dave Rust, I'm the Executive Secretary of the Agency, and since August, I've been the

Acting Deputy Commissioner for Disability and Income Security Programs. That's the policy shop in Social Security.

And I look forward to hearing the testimony today, and we're going to work closely with the Commissioner on converting the information we receive today into the -- the -- the policy framework needed to implement this initiative.

COMMISSIONER ASTRUE: Thank you.

Over the course of the day, we're going to examine how compassionate allowances can be implemented for individuals with rare diseases. This examination will include the perspectives of the advocacy, adjudicatory and medical technology community.

My thanks again to everyone for coming to today's public hearing. Whether you're here in person or listening in over the phone, perhaps with difficulty, I gather, I invite you to submit your thoughts about today's proceedings to the compassionate allowances mailbox at

compassionate.allowances@ssa.gov. Let me do that again: compassionate.allowances@ssa.gov.

Again, we appreciate your choosing to be a part of this process toward improving what we do in the disability area.

I'd now like to invite the first panel, which will provide an overview of rare diseases, to step to the table and kick off our hearing.

Let me welcome Diane Dorman, Paul Lipkin and Sissi Langford.

Diane Dorman is Vice President for Public Policy for the National Organization for Rare Disorders.

Paul Lipkin -- Dr. Paul Lipkin directs the Center for Development and Learning at the Kennedy Krieger Institute and is an Assistant Professor of Pediatrics at the Johns Hopkins University School of Medicine. He is testifying today on behalf of the American Academy of Pediatrics.

And Noni Langford, Chair of the

Committee on Federal Legislation for the National MPS Society, and is a caregiver for her two children with rare diseases.

Thank you all. Welcome.

We will begin with Diane.

MS. DORMAN: Good morning, Commissioner. My name is Diane Dorman, Vice President for Public Policy for the National Organization for Rare Disorders.

And on behalf of NORD and the millions of Americans affected by rare diseases, I want to thank you for the opportunity to speak before you today regarding the Social Security Administration's desire to improve the rules for compassionate allowances for individuals affected by rare diseases.

As mentioned in our written comments submitted to the Social Security Administration this past September, the men, women and children seriously affected by rare diseases, many of which are severely debilitating and are life-threatening, are routinely denied

Social Security disability insurance and are forced to go through the lengthy and often expensive appeals process.

This is because nearly 100 percent of the rare diseases are not in the SSA listing of impairments, nor are they included in the UN's International Classification of Diseases.

In many cases, initial denials of benefits is reversed following appeals, but not before patients and their families have lost precious time and spent thousands of dollars on legal assistance.

Rather than discussing the course of individual rare diseases, for the most part, I would like to provide a panoramic, kind of a 10,000-foot view of -- of rare diseases.

Now, according to the Office of Rare Diseases at NIH, there are approximately 7,000 known rare diseases, each of which affect fewer than 200,000 people in the U.S.

The NIH estimates that in the aggregate, between 9 and 10 percent of the U.S.

population has been diagnosed with one of these rare diseases, disorders or syndromes.

Eighty-five to 90 percent of known rare diseases are chronic, serious and/or life-threatening. Approximately 80 percent of these diseases are genetic.

Consequently, I make the assumption that children are inordinately impacted by these diseases. This is evidenced by the fact that about 50 percent of the over 300 orphan products are approved for pediatric use.

The diagnosis of a rare disease often takes years. Little evidence is available to support a diagnosis. Experts in the field are few and far between, and doctors know little of these diseases. As a result, patients and their families are shuffled from one specialist to another.

In a survey NORD conducted with Sarah Lawrence College several years ago, 42 percent of the respondents said that they were prevented from working because of their disease.

The survey was sent to about 15,000 people, with a response rate of 9 percent. In the same survey, 70 percent -- 77 percent of the respondents said they -- their rare disease had caused them or their families a financial burden. Thirty-two percent characterized the burden as extreme.

Many rare disorders have no surrogate endpoints, markers or tests. Some are diagnosed based on clinical observations only, other -- others by genetic tests.

Before closing, I would like to share with you one of the difficulties that have been described by -- described to me by one of the leaders of NORD's member organizations.

I will focus only on Marfan's disease -- syndrome at the moment, and additional information is available in my written comments about other diseases.

On November 3rd, 2004, another member of our group lost her fight with Marfan's syndrome, and believe me, she fought

hard. She was 39 years old.

She has spent the months since February 2004 trying to prepare her son for her ultimate death, make arrangements for him to be taken care of after her death, and try to get Social Security disability so she could take care of him while she lived. Just last week, Social Security denied her for the second time.

Honest to Pete, I don't know how they could do that. If they read her medical records and learned she still had a dissection that was not repaired at the time of the emergency surgery, that she could not walk even 15 feet without becoming totally out of breath, that she bled into her right leg if she stood too long, and she had suffered a heart attack, and she had Marfan's syndrome, how could they deny her?

Please, someone explain to me how the blazes they could turn her down. This woman had no means to provide for her son and herself except for the goodness of other people. She tried to work and just could not.

Up until the time she had her emergency dissection, she worked manual labor jobs, such as handling -- hand loading semis at a distribution warehouse to take care of her and her son. She did this because no one ever told her that she should not lift heavy things, especially not repetitive lifts.

Now I hope her family sends her obituary to the person who wrote the denial and ask them what might have been done had they -- might have been had they missed her request for help. It just does not seem that our system of helping people is working.

This is just one example of the types of frustrations, delays and unnecessary expenses often experienced by people with serious and even life-threatening diseases who apply for disability assistance. Several other examples are available in my testimony -- my written testimony.

When Commissioner Astrue spoke at the NORD annual conference in September, he emphasized that the impetus for change comes from SSA and not

from external sources; in other words, SSA truly desires to provide better service to claimants who are currently being subjected to unnecessary, emotionally draining and costly delays.

NORD applauds this proactive approach and wants to assure all involved that we and our medical advisors will support SSA in the effort to improve its service for patients with rare diseases in any way that we can.

Thank you, Commissioner. And I'd also like to extend a very special thanks to Diane Braunstein and Mary Shatel.

Thank you.

COMMISSIONER ASTRUE: Thank you, Diane.

I think what we're going to do here -- I think we may have been unclear on the procedure -- I think we're going to try to discipline ourselves and hold our questions until all three panelists have testified. Because I think when we have our questions, my guess is that two or three of you are going to want to comment

on -- on many of our questions, so we're going to hold our questions till the end.

And we'll move now to Dr. Lipkin.

DR. LIPKIN: Thank you, Commissioner, and good morning.

Again, my name is Paul Lipkin. I'm an assistant professor of pediatrics and a developmental pediatrician specializing in the care of children with disabilities at the Johns Hopkins University School of Medicine.

There I'm director of the Center for Development and Learning at the Kennedy Krieger Institute, which is a university center for excellence in developmental disabilities that's completely dedicated to the care of children with -- with disabilities and related medical conditions.

I'm also speaking as Immediate Past Chair of the American Academy of Pediatrics' Council on Children With Disabilities.

As someone who takes care of children who are eligibility -- who are eligible for

disability benefits from Social Security on a daily basis, I come today with firsthand experience knowing the struggles families can face when accessing benefits.

This includes prolonged waits for determination involving extensive information-gathering, examination, and for the families, uncertainty regarding their child and his or her future.

This is just one more burden for families who must struggle with a child's health, education and therapies every day.

I look forward to hearing Sissi Langford's experience as a parent working with her pediatrician and with Social Security.

I -- I made a point today of wearing this particular tie. It was designed by a 13-year-old -- now a 13-year-old child who I've known since approximately the age of 3. Emily is a happy, smiling 13-year-old child who, in the course of her 13 years of life, has had 35 neurosurgical procedures for some of the

congenital brain abnormalities.

So this is, for me, a reminder of the children that -- that I'm representing here today.

The 60,000 members of the American Academy of Pediatrics are on the front lines with families in diagnosing and treating children with rare diseases and other disabilities that limit their ability to function and develop in the same manner as their peers.

The Academy has had a long history of identifying and caring for children and youth with special healthcare needs, and of supporting the SSI program as a way to help children and families with resources and access to needed medical coverage through Medicaid.

As an example, I bring this booklet that the Academy has put together with the Social Security Administration on understanding SSI eligibility for children, and that's a vital tool for pediatricians in the process.

We've also authored policy statements specifically geared towards pediatricians to make

them better understand and to support the SSI program for families and -- and children that they are working with.

We are actually in the midst of revising this policy, and hopefully, in 2008, we'll see new support for pediatricians in this regard.

For a family with a child facing the need for increased healthcare services, frequent medical appointments and tests, prescription medications, medical devices, structured educational services and social supports, the resources provided through the SSI program and its linkage to Medicaid could be an enormous benefit. Indeed, it can make the differences for families and preserve the ability to remain in care.

The first thing that a child diagnosed with a rare disease or other severe medical condition needs is access to healthcare in a medical home.

The medical home is our approach to primary healthcare that ensures that children's

healthcare services are comprehensive, high quality and coordinated with community services.

In fact, the medical home model was first developed as a model for children like those we are talking about today, children with special healthcare needs.

For all -- for all children, but especially children with disabilities, coordinated care that connects them to all necessary health and community resources through a medical home is essential to maximizing their health and functioning.

The medical home is also an important linkage between families and Social Security. Pediatricians play an important role in identification, encouraging families to apply for benefits on behalf of their children.

They also provide the evidence Social Security needs to determine eligibility and advocate for families during the determination process.

The pediatrician can provide the

critical information needed regarding how a child lives every day with his or her rare disorder and her disability, and how he or she best relates to their peers and to the people that they relate to every day in school.

Enhancing the relationship between Social Security and a child's medical home should be an important part of any policy change Social Security's considering to streamline the application process.

Beyond the diseases and conditions already in the published medical criteria used by Social Security to determine eligibility, it's challenging to identify the additional number of children that could be made eligible under a new category of compassionate allowance.

We know that children with special healthcare needs make up approximately 12.8 percent, or 9.4 million children and youth in the United States.

This means that approximately one out of every five homes in the United States has a

child or youth with special healthcare needs living there.

This does not mean that these children are now or will ever be disabled according to the Social Security's standard of severity and duration as currently outlined.

Similarly, a child diagnosed with a rare disease or disorder may have immediate functional deficits, no functional deficits or may be at risk of diminished functional capacity as they age.

The critical issue from the pediatrician's perspective is that processing of an SSI application should be as expeditious as possible, linked closely with the child's medical home.

Care should be taken to ensure that only the minimal numbers of duplicate medical appointments or diagnostic tests necessary are required.

Children and families with disabilities, including rare disorders, face an

array of complex and demanding network of healthcare providers and community resources. A medical home can help coordinate and streamline this to the extent possible.

Social Security should be willing to work with the medical home to gather needed medical evidence, thereby reducing the burden on children and families.

I believe we're on the cusp of an important new era in healthcare for children. We now can better identify children with rare disorders with exotic names such as mucopolysaccharidosis, ornithine transcarbamylase deficiency, Gaucher's Disease.

Thanks to the advances in neuroscience and genetics, we can now identify these children earlier, provide new, all -- albeit expensive, specialized medical treatments, and thus improve their short- and long-term health, social integration and daily functioning in ways not previously available.

We also expect these advances to

continue in the coming decades.

The Compassionate Allowance Program being considered will spare these families prolonged uncertainty and unnecessary hardship and delay.

We look forward to your consideration of these factors.

Thank you for the opportunity to address these issues. I look forward to answering your questions.

COMMISSIONER ASTRUE: Thank you.

Thank you. I -- I -- I've got an update that the phone line apparently is working well, but I -- I've been asked to stress to speak as loudly as you can, as close to the microphone as possible. And I'll try to live up to my own -- my own guidance, too.

Thanks.

Sissi.

MS. LANGFORD: Sure. Thank you, Social Security Commissioner Astrue, for this opportunity to represent the National MPS Society

and to talk with you about my family and our experiences navigating the disability determination approval process for Social Security.

Thanks for making me feel so welcome here today.

I feel strongly that children with MPS can teach all of us the important role the Government plays in caring for people with severe disabilities, and I appreciate this opportunity to share my thoughts with you.

My name is Sissi Langford. I am an elected board member of the National MPS Society. I chair the Committee on Federal Legislation. My committee acts as a liaison between Congress and Government agencies and our general membership.

We -- the National MPS Society is a nonprofit 501(c)(3) family support organization. Our goal is to ultimately find a cure for -- for MPS disorders by supporting research and providing support to -- to individuals and their families affected by MPS and, more importantly, by

promoting public and professional awareness of these disorders.

Due to the often short-life expectancy of people affected with MPS, the majority of our affected members are children and their families.

Our membership does also include a small percentage of adults who suffer from MPS.

Some children are considered mildly affected, while others seek treatments which have drastically approved their qualities of life.

Some examples of new treatments are the enzyme replacement therapy and the bone marrow transplant and cord blood stem cell transplant.

These have made tremendous differences in the lives of these children. These -- these treatments are not cures, and they're not available for all MPS children.

I recognize that these factors make it difficult to set policy for the Social Security Administration benefit approval process.

The mucopolysaccharidosis, or MPS, disorders are genetically determined lysosomal

storage diseases. They result in a body's inability to break down enzymes. Basically, the enzymes are stored within the cells in the body, and over time, they damage these cells and eventually destroy the cells.

Storage causes progressive cell damage in multiple systems within the body, including respiratory, bones, internal organs, heart and central nervous system and significant cognitive -- cognitive delay.

The results of the damages, it includes mental retardation, short stature, corneal damage, joint stiffness, loss of mobility, speech and hearing impairment, heart disease, hyperactivity, chronic respiratory and digestive problems, and most importantly, drastically shortened life spans.

My 12-year-old son, Joe, and my 11-year-old daughter, Maggie, suffer from Sanfilippo syndrome, or MPS III. It is a result of a genetic recessive mutation that both -- in both mine and my husband's DNA. There's a

one-in-four chance that each one of our children will have a mutation that causes MPS III.

Because of the extensive central nervous system involvement, there are currently no treatments available for children with MPS III.

We live on Johns Island in South Carolina near Charleston, where we receive very good state and county level services and have access to a medical university for the management of this disorder.

I've learned that many other families do not have the same level of care we have available in South Carolina and do not experience the -- the same level of success we have had with getting benefits for these children.

Joe was born in February 1995 and developed normally, except for frequent ear infections and frequent diarrhea. Maggie was born in August 1996 -- they're 19 months apart -- and also appeared to be a normal, healthy child.

Joe started dropping words out of his vocabulary at around age 3, at a time when he

should have been gaining words. And our pediatrician noticed that his spleen was enlarged, which was an indication of a storage problem.

Our pediatrician referred Joe to a geneticist for testing and to the BabyNet program in May of 1998.

The diagnosis process took over a year, and during that time, we realized that Maggie was experiencing similar decline.

Joe and Maggie were officially diagnosed with Sanfilippo syndrome in June 1999.

My husband and I researched these MPS disorders, and we were devastated by what we learned. Joe and Maggie seemed mostly normal, so we could not imagine the decline that we -- that we read as described in the -- in the research.

Now, Joe and Maggie depend on wheelchairs completely for mobility. They are no longer verbal and have lost their ability to swallow. They do not laugh or cry. They both rely completely on feeding tubes for nutrition. They have heart defects, and they're treated for

seizures. They also take medications for sleep and to control agitation and involuntary movement.

They're seen regularly by cardiologists, neurologists, gastroenterologists, orthopedists and other specialists.

Joe and Maggie are now developmentally about 12 months old because of those significant cognitive involvement from MPS III. They will continue to decline as MPS causes more medical problems.

Children with MPS III have a life expectancy of an average of 14 years. It is our hope that Joe and Maggie will live longer than average and that they can continue to enjoy an acceptable quality of life.

It is an overwhelming challenge to provide our children with an acceptable quality of life due to their decline and the current level of care required to keep them as healthy as possible.

Our family's ability to manage the care of Joe and Maggie is a direct result of the services we -- we receive from the State of

Cal- -- South Carolina, which includes funding from the Federal Government.

The process begins with the Social Security Administration's determination of eligibility. Many families fail to realize the importance of determining eligibility early in their child's lives.

MPS disorders are all degenerative, which makes it difficult for parents, teachers and medical providers to accept that an active, vocal, mobile child will lose these skills.

My husband and I were reluctant to apply to the Social Security Administration for the disability determination, because we had not accepted the reality of our children's condition, and because we felt we should be able to handle our family's medical needs with our private insurance and our personal resources.

The early interventionalists and other health professionals working with us recognized that we struggled with this diagnosis, and they consistently persuaded us to prepare for the

future.

We were strongly advised to pursue any services available to our family. The process is -- the process was long and complicated.

While Joe was accepted through the SSA for Medicaid and the waiver, Maggie was declined because she was not delayed enough. I had to appeal this decision and attended a hearing in order to get Maggie on the waiting list for services.

I had to learn my rights and how to get my doctors to write meaningful letters to describe our situation. This took several years.

Our local agencies recommended other funding programs and encouraged us to apply for grants to pay for diapers, which were over $350 a month, during this long delay to get through the approval process and off the waiting lists.

I've included a description of those State-provided services and budget for Joe and Maggie just to give you an idea of -- of what -- how it looks from our point.

I'm sure you -- you guys already know most of this, but Joe and Maggie are covered under the TEFRA, or Katie Beckett Waiver, which is a category for children under the age of 19 and is designed to keep the child in the home instead of in the community or hospital setting.

We applied for the TEFRA program for both kids in October of '99. Joe was made eligible about a month later, in November of '99, and was placed on a waiting list in slot Number 12.

Maggie was denied that eligibility in May 2000, and the hearing to appeal the denial was in August of 2000. At that point, Maggie was ruled eligible.

Both children began getting services under MR/MD Waiver in February 2003. So this process took from October '99 to February 2003.

Joe's estimate -- estimated budget for the 2006-2007 year is $69,000, and Maggie's is 79,000. That does not reflect the amount of money our private insurance pays out, which is

80 percent of the -- of the billing for medical needs.

The children are -- are eligible for additional nursing hours, but it's hard to find more nursing care at this time.

The budget reflects the nursing care and personal care assistants we use, not the total that they are eligible for.

But even at this extensive level of assistance, we have completely altered our life styles to care for our children, including moving into a new home near a school that was appropriate for them. I can no longer work outside my home, and I've had to drastically reduce my professional workload to manage the care of my children.

I'm a residential designer. I'm fortunate that I can work from my home, but I've had to reduce my ability to expand my services to my clients.

We will continue to do what we can to make sure Joe and Maggie are as healthy as they can be as they progress and eventually become

unable to keep up with the devastating effects of MPS.

It is impossible for me to express how hard these children fight for their health. Their ability to keep up this fight has given me the courage and the inspiration to try to seek improvements for all MPS children and for rare disease groups in general.

The following points are -- are relevant from the South Carolina Department of Disabilities, just to give you a little picture of -- of what it is like where I live.

They receive about $300 million in Federal Medicaid funding to provide services. They currently serve 28,000 people. Approximately 82 percent of these people live at home with their families, compared to only 60 percent nationally.

The -- the report that I got this information from lists the following opportunities: to increase the use of Medicaid funding to provide flexible in-home support for increased individual/family and dependents, and to

strengthen technology capabilities, and to enhance service provider productivity and deficiency.

They also pointed out a few barriers, and -- and mostly the barriers have to do with the long waiting lists. When my son was Number 12 on the waiting list, that didn't seem so bad. I didn't know it would be three years later before he was at the top of the list.

And then their other barrier is, again, the recruitment and the retention of nurses. So that is -- this continues to be a big problem.

We have a matrix of caregivers who come into our home. We don't have people all the time, but we have three nights a week covered, and we have an LPN during the day who goes to school with our kids to handle their meds and their feeds.

It is encouraging to know that my state recognizes some of the same challenges that parents see, and it is my hope that South Carolina and other States will continue to look for

solutions.

It is also encourage -- encouraging that Commissioner Astrue is committed to improving the disability determination process for the disability insurance and supplemental security income programs.

I think it's important to -- to really stress the -- the -- the problem from a point of view of a parent. It is -- the -- many parents have expressed that the lack of knowledge about MPS is -- is the biggest problem, specifically, the degree and the rate of decline that these children face. Most parents eventually recognize that they must advocate for their child and that they must educate everyone involved with their child's care about the nature of MPS.

The disorder is rare, and it's hard to understand. It's hard to pronounce, it's hard to spell, and it's hard to remember unless you have a child with it. It is degenerative, and it is impossible to predict how quickly decline will occur.

Unfortunately, many parents are not emotionally ready to take on the role of advocate soon after diagnosis when they should be pursuing these approval processes.

If MPS was on the compassionate allowance list, it would certainly lessen that burden on the parent. It would allow the state to start helping the family as early as possible, which would give the parents some time to acclimate to the important role of advocate.

Early approval -- it's impart -- important to recognize, too, that success depends on the approval of services prior to need. In our case, we -- because we had -- we struggled so much with this diagnosis and this decline, we had some strong support.

Our physical therapist, for one, just insisted that I pursue getting an adaptive van with modifications to be able to transfer my children. My children were still basically walking with help at that point. But she -- she continued to harass me to do this, and finally I

did it so that by the time my children were completely confined to their chairs, we had this important milestone marked off our list.

Now we can safely transport our children to doctors' appointments and -- and wherever they need to go.

But our success was a result of an experienced professional who could predict our need and who went beyond their required scope of her job as a physical therapist to get me on board.

Most parents are not that lucky. I was fortunate that I have people who -- who were able to guide me through this.

Another problem that MPS parents have experienced is a lack of technical training for service coordinators who must seek approvals for services.

Parents state that the service coordinators and case workers are not adequately trained to file paperwork and -- that would not be denied, or the approving agency is not trained to

respond so problems can be resolved quickly.

Parents request that this type of delay in approval of services be addressed by providing relevant, regular technical training for everyone involved in the approval process.

And then I think it's important to identify strong service coordinators. It's not practical for all service coordinators to be trained in the unique challenges of rare disorders.

Identify and match strong service coordinators with families with rare diseases. Our service coordinator is a -- a -- a very vital part of our picture. She -- she predicts our needs ahead of time. And most importantly, she writes letters and assists medical professionals with writing letters that get approved.

I've had several other service coordinators who had different and more unsuccessful approaches.

I had to learn to advocate for a service coordinator who could handle our

situation.

MPS parents and Government agencies share the same goal: keep children at home while providing support that allows the family to continue to function.

The parent and the service coordinator must develop a working relationship to plan ahead for the challenges of caring for these MPS children. The doctors and other health professionals must be open to writing descriptive letters that illustrate the disease process and allow the reviewer to understand the complex nature of MPS.

The approving agencies must recognize that there's not a cookbook approach to approving the services needed to keep these children in their homes.

Information describing the impact of the disorder would be helpful, but a change in the philosophy for handling a rare degenerative disorder like MPS will provide the greatest improvement.

Thank you for taking time to learn about MPS and Joe and Maggie. And I appreciate the opportunity to share this with you and the fact that you value my views. I think this is an important step to improving the approval process.

COMMISSIONER ASTRUE: Thank you very much.

Let -- let me kick off the first question. It will be a long-winded one, and I apologize for that. And it's going to eventually loop around the question, how can we do what we need to do quickly, consistent with doing it well.

We have a number of constraints in this process. We have a statute that says we can only give disability for people that we believe are unable to work for 12 months or more. This is shorthand for it.

So, for instance, one of the things that means for us is that for some degenerative diseases, at initial diagnosis, the person doesn't meet our standards, but we know with certainty that they will later on. And so we need to come

up with standards and markers to try to draw that line in an appropriate way. And sometimes genetic information helps, but sometimes it makes it harder.

You know, for instance, with Huntington's disease, it used to be probably at the point that you -- they were eventually diagnosed, those patients, you -- you knew that they were disabled under our standards. Now they can be diagnosed at birth with genetic, and they may not reach submit -- may not have significant impairments until the time they're 40.

So we have a statutory obligation to balance that out to draw those kinds of lines, which makes the task of going through all 7,000 rare diseases to try to figure out what to say about them very difficult. And we've got now more options in terms of what we can say.

We can do the traditional route and just simply put guidance into the listings. We can put something in the quick disability determination list and say it's highly probable,

but not certain, where we still go through our -- our normal processes, we just move it up to the top of the queue, and so they get decided very quickly. Or we can, and in probably what will be a relatively limited number of cases, go to conclusive presumptions and call something a "compassionate allowance."

Operationally, this is an enormous task for us. I think, if I remember correctly, David has about 150 people to do all the medical-related work, and you start looking at the size of what they've already done in the past, asking them to do a lot more, you could easily see how that could bog down and get slow.

So we talked about -- we've been doing some talking about how do we go about, just from an operational point of view, doing this well and quickly. You know, we've talked about perhaps hiring contractors to assist us to go through this list, and it might be helpful to know what types of organizations might be able to do this work, have the adequate medical knowledge, but also

apply our statutory standard to it to make sure that we -- we get what we need, the right end result at the end.

We've reached out, obviously, to NIH and to NORD and lots of other organizations, but I think that task probably isn't finished yet, and we probably haven't talked to all the organizations that might have information that could help us do this well and quickly. And then when we've gone through the first time through, to update on a regular basis.

So I just thought I'd -- I'd throw it open for initial question to see if you have any advice or guidance -- if you were in our shoes or, more accurately, in David's shoes, since he's on the front line on this, how would you think about this, how would you organize the task, because, you know, you look at -- you spend a lot of time looking at one disease, it all becomes very obvious. But if you -- you look at a relatively small organization that's trying to look at a ton of these, how do you do that efficiently and well?

Any -- any suggestions or guidance?

MS. DORMAN: First of all, I mean, you've made a really good start.

COMMISSIONER ASTRUE: Thank you.

MS. DORMAN: I mean, you're -- you're out there reaching out to Steve Groft's office at the NIH, to NORD, or the patient organizations, American Academy of Pediatrics.

So I think you've made a really important first step. And I think possibly identifying maybe -- I hate using this phrase, but the low lying fruit, so to speak, and turning to experts, NORD's medical advisory committee, the medical advisory committees of other organizations working in conjunction with -- with Steve Groft's office.

I think that is probably one of the most important things that you've done so far. I don't have the answers to the really bigger questions, but I think you've made a really important step forward.

DR. LIPKIN: It's an enormous task,

when you talk about 7 -- 7,000 diseases. And many of them, there may be only a handful of people in the country who really have expertise in that particular disease.

It -- it seems to me as if some sort of consultative panel of -- of experts needs -- would be useful in terms of convening to consider the 7,000 disorders in one way or another, people with specialized expertise, not necessarily on one particular disorder, but on these classes of problems. And, of course, not only the medical expertise that may be necessary, but -- but people with personal and -- and the social experience to go through that as well.

But I think some sort of a panel is probably going to be imperative, who can provide you with some sort of report that you could work from.

MS. DORMAN: I would like to say maybe, Steve, it's time to bring together the panel and kind of relook at that report that was issued by the Commission back in 1989. I know

that's a big task, but I think that would probably be -- since the model is already available and -- and -- and worked, that would probably be the most viable opportunity for -- for that suggestion.

DR. GROFT: I -- I think that's part of our involvement here is -- is we're going to look at -- at what is available, the resources that are available, especially in -- in the medical profession, the specialty societies that -- that will be necessary to make those judgments.

And -- and I think it's important, too, to understand, too, that many of the patient advocacy groups have outstanding medical and scientific advisory boards that we shouldn't overlook.

And -- and, Sissi, if I can just ask a question, too --

MS. LANGFORD: Sure.

DR. GROFT: -- the -- the willingness of -- of your medical advisory board to participate in such an activity of maybe saying

when does disability really start to occur, is -- is there a level that we can say is a -- a dropoff point for the patients that we really need to be concerned about? Have you discussed this with your board?

MS. LANGFORD: I have. Unfortunately, because of the -- the way the diseases appear, it -- it varies greatly. You know, even within our family, it -- it would -- it would have been hard for a pediatrician to -- to -- to know that these -- both these kids had the same disease. It took a long time and just -- we just started sensing that Maggie had what Joe had.

I -- I think one positive thing, though, about the way our medical advisory boards work, is, in the rare diseases, chances are the -- the doctors on our -- our board are also on some other boards, the Batten Disease and some other related lysosomal storage diseases.

So the -- the community grows as the researchers do research on -- on different diseases and work with those organizations, so

some of these -- these researchers have a very good understanding of many of the -- of a type of disease, so they -- they can -- they can offer that.

But, again, I mean, any time we even asked, you know, our scientific -- you know, my geneticist, who's of course on our board, I asked them questions about things that are happening with my children, and they -- they're not sure if -- you know, if that's related or if it's -- you know, they're not -- there's just -- it's just impossible to predict the outcome of the disease.

DR. LIPKIN: You know, I -- I think we're asking the Administration and -- and whatever panel were -- were to be put together, to -- you know, of course, to play the role of King Solomon in some way and -- and -- and make the difficult choices.

The -- on the other hand, I think there are certain conditions that are clearly more devastating than others and really probably do

need to rise to the top and do need to be considered first, within the system.

So although, we're talking about 7,000, there is going to be a core that is -- that is absolutely devastating and -- and -- and heartbreaking in many ways and that are probably not going to be quite as difficult to make decisions about.

COMMISSIONER ASTRUE: Dave, do you have any?

ACTING DEPUTY COMMISSIONER RUST: Doctor -- Dr. Lipkin, one of the things that --

UNIDENTIFIABLE VOICE: We can't hear you.

COMMISSIONER ASTRUE: You get very -- you've got to get very close to the microphone or we're -- we're going to end up talking to someone we can't see again.

ACTING DEPUTY COMMISSIONER RUST: Dr. Lipkin, one of the -- I thought one of the interesting observations that Ms. Langford made was the fact that the doctors did not know how to

present the information to the disability process. They're thinking as physicians, they're thinking in terms of treating the patient; they're not thinking in terms of translating that into the information the examiner needs.

Do you have any suggestions on how the -- the physicians that treat these rare diseases might be, you know, give -- given information that would help them to provide more useful information to the disability process?

DR. LIPKIN: Yeah. Well, certainly the -- the physician is confronted with forms that can be sometimes lengthy, obtuse, and -- and not in a language that -- that he or she is -- is familiar with in terms of thinking, certainly has not been trained to complete.

The -- certainly the -- I think the -- the -- the specialists who are dealing with some of these disorders have had more of a routine of being asked for such information, but nevertheless, I think often will rely upon administrative staff and support people to do

that.

I think -- I think there probably does need to be consideration for some core information, particularly when we're talking about rare disorders, and some key questions that are in -- in -- in common medical language and medical terminology that will be important.

I think at the same time, I think in -- in medicine, we are moving more and more towards this concept of functional skills, functional impairments. Quite frankly, I think most physicians, including pediatricians, that's still a foreign concept. Hopefully, newer people coming to the field of medicine will better understand them.

But I think that is a language that both the medical field and Social Security needs to adapt, because I think it's -- it's completely intended for these purposes. And so there probably needs to be a joint educational process and -- and any -- and any inquiries that -- that are requested from Social Security office probably

needs to use that -- those new approaches.

ACTING DEPUTY COMMISSIONER RUST: Thank you.

MS. LANGFORD: I also feel it would be beneficial if the doctors would take advantage of the -- the support groups, because we -- we certainly provide a lot of literature, and we can connect doctors with researchers who are familiar and who actually, like in England, where clinicians who see Sanfilippo children their whole lives because of the way their government health works there.

So there -- there is a lot of information available that's at that doctor level, and -- and I think it -- as a parent, it's my responsibility to encourage my doctor to seek that out.

It's -- it's hard -- it's a hard -- it can be a hard sell to get the doctors to want to do that, and I -- and I recognize that. They have -- you know, they have a lot to do. But there needs to be some way to get them used to

reaching out to the family support groups.

We've tried to go to the doctor, you know, committee -- you know, large meeting conferences and stuff, and there's so -- there's such an overload of information there, it's probably not beneficial to hand out our brochures at, you know, American Academy of Pediatrics.

DR. LIPKIN: Right. You know, Ms. Langford points out some -- some important issues in terms of information gaps that -- that physicians will have. I -- I think it probably is worthwhile highlighting here that in nearly every state in the union, there are Federally funded UCETs, or university centers, that are intended to provide assistance for families, both medically and therapeutically.

And, quite frankly, I think many -- a few physicians even know about the existence of such facilities. And I think they are probably going to be important key players in assisting this process as well.

MS. DORMAN: And it -- and it's just

not the responsibility of the Social Security Administration, it's the -- it's the entire rare disease community working interactively with one another.

I was looking at this particular rare disease, which I'm going to try to pronounce, I'm going to break it down, lymphangioleiomyomatosis. So if I were a doctor or if I were a reviewer at the Social Security Administration and I saw that, I would have no clue whatsoever about what to do about that particular rare disease.

So I think it's communicating, reaching out, and all working with one another, because, although the community in the aggregate is very large, in essence, it's very, very small, and we need to be reaching out to one another more.

COMMISSIONER ASTRUE: It -- it -- it's -- it's a -- it's an outstanding point.

Let me just talk to that briefly, and I want to make sure I give Frank a -- a chance, too, I don't want to hog the forum.

But one of the ways to conceptualize the problem that we have here is it's -- it's just a lack of information. And one of the things I think it's important to understand is how the process works. It's by statute, the States make the first two levels of decisionmaking for us.

And we have representatives, I know, from the National Association of Disability Examiners here. And it would be a good idea in the break, for instance, to corner some of them and talk to them about what they do and how they do it.

And I talk -- I talk to a lot of these folks. They're very low paid compared to Federal standards. The turnover is very high. In many States, the average experience of an examiner is about three years. And, of course, medical school takes four. And these are -- tend to be, almost without exception, terrific people. They self-select because they want to help. And they often have a social work degree, a psychology degree. They're terrific people.

But you look at the vast array of things here, they get very good, very quickly at the cases they see all the time. And then they turn to experts, and they have in-house experts, medical experts, for the diseases that they don't recognize. But there the problem is, one, where, again, we don't have rare disease experts, it would be impractical to do that actually working within the DDSs, and I think what typically happens is that a generalist dusts off his medical textbooks, and there isn't very much there, either.

I remember there was a stunning case, I -- I remember talking to the head of the MPS society for Europe, and I think it was mucopolysaccharidosis to Hunter disease, and the -- the -- and I may have that wrong, I may have the disease wrong, but the -- a -- a mom applied for disability for the child. And they have a similar system in the UK, and the doc went to the textbook, and the disease wasn't there.

And so he essentially -- the -- the --

the local disability arm actually started prosecuting the family for fraud, you know, and said the disease didn't exist or the symptoms didn't exist and that type of thing.

So it -- it -- it is important -- it's important, while it's -- it's very natural when you're frustrated to assume that there've got to be badly motivated people in the process, all -- almost without exception, that's not the case. These are absolutely terrific people who work in very difficult circumstances.

And it -- it is -- a lot of the -- the -- the problems aren't their fault, they're our fault, because we need to make it easier when they see a disease that they haven't seen before, to click on their increasingly sophisticated computers and punch in Marfan's or punch in Sanfilippo and have some form of guidance, and whether it's in the listings, whether it's QDD or what it is, I think that's really what we need to do.

And it's important not just for the

examiners, but it's important because the physicians often have those gaps, too, and it's because the textbooks and a lot of the standard things that they look for tend to overlook the rare diseases.

So I think the way to conceptualize is, a lot of the times, is how do we get the right information easily to the people that make the -- the decisions. And to the extent you can help us with that, because a lot of these diseases, again, it's going to be -- we can't put by statute everybody in, and we're going to have to figure out how to draw lines, and to the extent that particularly the physicians that work in your areas can help us draw those lines intelligently and consistent with the statute, that's going to help us move along this project much, much faster if we can do that.

MS. DORMAN: My mother gave me some really good advice as I was growing up. She said, "you don't have to know everything, just hang around with really smart people." And so I think

that's really important, a very good lesson that we all have to -- to abide by is -- is working with one another.

COMMISSIONER ASTRUE: Yeah. That's right. Well, as an English major in biotech, I learned that pretty -- pretty darn fast.

Frank, do you have some questions?

JUDGE CRISTAUDO: Thank you. Follow up -- I would like to follow up with the point that the Commissioner just made --

COMMISSIONER ASTRUE: Move a little closer to the -- bring your mike a little closer.

JUDGE CRISTAUDO: I would like to follow up with the point that the Commissioner just made and that David asked about. Certainly the information that the medical sources provide to us is obviously critical, and if you have any further thoughts on -- on how we can encourage or make that process easier so we get the information we need earlier, certainly it would be very helpful.

But it also goes to the -- the

families and the other people familiar with a particular situation. So if you have any thoughts on how -- how we can approach, perhaps, encouraging more of that information to be provided earlier in the process.

As I put it out, I'm -- I'm an administrative law judge. Many of the cases we see, we end up finding out that there's some additional evidence that was not provided earlier. So if you have any other thoughts on how we could improve that process, I certainly would like to hear that.

DR. LIPKIN: I think you -- I mean, you both nicely highlighted the importance of all the information that's now available online. And -- and so not only is that available to the people making disability determination, but to the pediatrician who has never heard of -- of XY or -- or Z disease and his textbook doesn't have it, or to the parent as well. And actually, the NIH has -- has done a great job with making information like that available; CDC has done that

as well.

And I think those are excellent vehicles that -- that all communities really take advantage of, and I think it's probably going to be critical -- critical information in this whole process.

COMMISSIONER ASTRUE: One -- one -- one of the things that maybe to think about, I know at the -- the risk -- we're moving this into more informal dialogue than -- than formal question and answer, but -- but that's okay as far as I'm concerned.

One of the things that if you're with a disease association you might think about doing is, I know a lot of times the Web sites have a lot of very helpful information often about how to apply for Medicare and -- and -- and, you know, where to find free drugs, and -- and they're a true -- terrific resource for the patients.

One of the things you may think about is helping yourselves to help us to the extent that you can come up with explanations of what our

process requires. In -- in particular, what you need to tell us in terms of functionality that's important for the particular disease, that might be very helpful to patients if we do have to make an ad hoc decision.

The other thing -- and -- and this is a very practical thing, and it's a great frustration for a lot of the people who are in the system. One of the reasons we make -- one of the -- the big reasons we make a lot of incorrect decisions is that we just don't have the full medical record. And so one of the things that's really important to stress to your members is to get the complete medical record in as early in the process as possible.

A lot of times we actually don't see the full medical record until the -- the case has already been kicking around for a couple of years. And that's just -- that's bad for you, that's bad for us, that's bad for everybody.

So, you know, mostly, you know, we're trying to figure out how to do our own work

better, but you may be also, many of you in the audience and many of you listening to this in remote locations, you may also be able to help your own members to work for us if they know kind of what our requirements are for applying for disability and what things we need to look at in terms of functionality.

And I think that might be -- be helpful to a lot of the people going through the process that perhaps aren't going to benefit from a compassionate allowance because it's one of those diseases where we do have to make an ad hoc decision.

But knowing more about our rules and what we need and what we need to look at might help people get through the process more quickly and -- and with -- with better results.

DR. LIPKIN: Certainly any fast-track system needs to be made available online and -- and certainly would expedite families' ability to apply, but also for the physician, as well, provide that key information. You know,

particularly if you're talking about getting medical records -- hard copies of medical records from -- from hospitals, you know, it could take months.

And -- and -- and -- and we're -- we're more moving towards electronic record systems as well. And -- and -- and pediatricians and family physicians in our offices are going to start having that more, and I think -- and I think we need to have a system for such expedited review that can really make that information available very quickly.

COMMISSIONER ASTRUE: That -- that's a terrific point that I actually didn't anticipate coming up today, but we are working on it. It's going to take us a while. Our system for online application is not as quick -- it's true for retirement as well as disability. And we have an ongoing effort to try to streamline those, make them user friendly. That's a pretty big effort in our world.

And we're also wrestling with how to

plug in medical records, because the -- the system is in -- in transition. And so, you know, I guess about a third of all Americans now have pretty complete electronic records.

So we're trying to figure out how to adapt our system so that we can take full advantage of that going forward. We're not there yet. But you're exactly right. We need to do that well in order to do this quickly.

DR. LIPKIN: Right.

COMMISSIONER ASTRUE: Any other questions?

ACTING DEPUTY COMMISSIONER RUST: No.

COMMISSIONER ASTRUE: You got another one?

JUDGE CRISTAUDO: Doctor, there's been a lot of discussion here about function, and we've obviously been asking questions about how we can improve the process of getting information as early as possible in the process.

One of -- one of the discussions that honestly I've -- I've thought some about is, is

when we ask physicians or medical sources for information about function, many times we're asking the medical sources doctor, what can this person do, despite their impairments?

And I've had some conversations with some medical sources where they suggest that maybe a better approach would be ask to something like, well, you know, as a result of this impairment, what is it the person cannot do? What are the limitations imposed by the impairment?

Do -- do med -- do physicians look at that differently, asking them what a -- a person can do versus what a person cannot do?

DR. LIPKIN: Absolutely. And particularly when you're talking about -- whether you're talking about children or adults, I think it is much easier information to glean and -- and, in fact, when the new classification system for function, international classification of function, ICF, that was put together internationally, they rely a -- a lot upon descriptions of children in multiple different

scenarios, adults as well in multiple different scenarios. Exactly how they handle being on the street, how they handle being in a supermarket, how they handle even mountain climbing, interestingly enough.

So -- so, absolutely. I think while -- while we've always functioned on disability and impairment, the ability is I -- is really, I guess, what we want to tap into realistically, what is this person able to do rather than unable to do.

JUDGE CRISTAUDO: The -- the question really goes to what -- what is easier for the -- for the physician to answer. Is -- is an easier question to answer what the person can do versus what they can't do or vice versa?

DR. LIPKIN: Yeah. I -- I think, well, the physician, whether it be interviewing the patient themselves or the parent, can -- certainly knows how to ask and can ask well as to what -- where that person goes every day and what they to. And -- and rather than thinking about

all the things they don't do, it's very easy for a family member to explain what a person does do and what their daily life is like.

JUDGE CRISTAUDO: Thank you.

MS. LANGFORD: I -- I'd like to add that I think it's relevant to say what can't be done in order to emphasize the need for the services.

The example is we -- our -- our children needed aides in school, one-on-one aides, because at a particular point in Sanfilippo, the children are extremely hyper, and they were -- they were damaging other kids, and they were in a disability class.

We had to write letters to get aides assigned to Joe and Maggie based on what they can't do, which is, you know, behave in class or whatever.

And so I think if we -- if -- as far as getting approval processes go -- going, I think it makes sense to -- to state in every day situation what the child can't do.

DR. GROFT: If I -- if I could just -- if I could just add that it would be important for us to hear of the -- the data elements or the criteria that would be necessary that you'd like to receive, so that perhaps we -- we need to repackage the information that is available to the public.

I -- I think if one looks at the -- what is made available through the patient advocacy groups, through the NIH, through NORD, other organizations, that there's considerable information available, but it's how -- how we package it doesn't really make sense for a reviewer or someone to look at, and perhaps you could help us identify what is actually needed to -- to facilitate your decisionmaking down at the -- at that -- that local level. So that -- that would be helpful.

And I know the discussions have been held with the Library of Medicine that the folks seem to be very willing to -- to -- to work with Social Security Administration to -- to provide

whatever we can do to -- to assist.

COMMISSIONER ASTRUE: Yeah. No, I know NIH has been absolutely spectacular so far, and we're very grateful.

Okay. I think despite our technological glitches, we're off to a great start. So I want to thank the witnesses. This has been tremendously helpful. We're grateful for your input.

And we're actually pretty much on time, and I -- and I think I can give you a decent break.

So why don't we -- why don't we take half an hour and come back at 11:10, and then we'll move to our next panel.

So thank you very much.

MS. DORMAN: Thank you, Commissioner.

(Recess.)

COMMISSIONER ASTRUE: We're going to try to make sure that -- we consider timeliness an important value of the agency.

We're going to try to get back on

track here and move to our next panel. So I know there are probably some good conversations going on, but I'd like to interrupt that so that we can give the panel our full attention.

So we have another three-person panel, very distinguished panel.

We have Linda Landry, who is a senior attorney from the Disability Law Center in Boston.

We have William Leach, who is a staff attorney for the A.C.C.E.S.S. Program in Tampa, as well as Kim Bernstein, who is director of the A.C.C.E.S.S. Program in Tampa.

Thank you very much for joining us today.

MS. LANDRY: Me first.

COMMISSIONER ASTRUE: Whichever way you want to go.

Yeah. And -- and -- and -- and I hate to be repetitive, and people have had to remind me, too, that because of our technological limitations in the room, speak as close to the microphone as you can. I realize it's a little

uncomfortable, and I apologize, but that's the way to make sure everybody hears what you have to say.

MS. LANDRY: Thank you very much. Thank you for this opportunity to testify.

As you said, my name is Linda Landry. I'm a senior attorney at the Disability Law Center. I've been representing people before the Social Security Administration for about 27 years.

And it's my opinion that the problems with disability determinations for people with rare conditions and diseases are pretty much the same as the problems with the process for -- for everybody else. It's perhaps more acute with people with rare conditions and -- and diseases where they're even less well-known than -- than other conditions.

But I think the problem starts right at the very beginning, with the application. Not enough of the right kind of information is identified, is requested of applicants when they apply. It's very medical/doctor/hospital specific.

And when you're talking about a lot of the disability adjudications that are needed, you also need other sources of information that -- that supply that critical functional capacity, ability to function on a day-to-day basis information. Often the doctors don't really have that, spend enough time with the -- with the individual who is applying.

And it is other resources out there, other nondoctor professional medical resources, other nonmedical professional sources; so if you're talking about children, it's school information, counselors and therapists, and that -- that -- those sources tend not to be identified. Unless the advocate, the -- the applicant has been sort of coached to provide that information, it's not really sought on the application.

And I also think that when sources are requested to provide information from Social Security, they're really not asked specifically enough for what Social Security

needs.

In Massachusetts, we work pretty closely with our DDS, which I think overall does -- does a really great job, and I agree with you entitle -- entirely, Commissioner, that they're underpaid and -- and overworked.

But the -- the -- they don't -- the -- some have -- some of the -- they have forms for some conditions and diseases, but certainly not all of them. And some of them are out of date, and we've been talking about trying to update them, but the time and the money doesn't seem to be available to do that.

I don't think doctors and other sources of information really understand the disability determination -- the disability definition for adults or children, and they don't really know how to respond.

And a -- and a -- when an advocate gets involved, that's a lot of what an advocate does is, so, you know, try to send them information that they'll read and are able to

digest about -- about what the disability standard is and what Social Security really needs to determine whether somebody, A, has a medically determinable impairment, and, B, what the nature and severity of that impairment is and -- and whether it actually means that an adult is unable to work for 12 months or a child has marked and severe limitations such that they should be considered eligible under the childhood disability standard.

You know, the other thing that happens in -- in evaluating information from a lot of these sources is, you know, you're getting the information, especially the readily prepared information, the already prepared information in the ordinary course that is -- they're talking to each other.

They use terms and phrases that -- that they know what it means, but it -- it doesn't necessarily correlate into the language of the disability standard, and sometimes it's like two foreign languages colliding.

You know, often I will see a -- a person denied because in the medical notes it says the individual is doing well, but it often means well under the circumstances. It doesn't mean they're better. It doesn't even necessarily mean they can function well enough to -- to work or -- or to function as a -- as a child without a disability would -- would function. You know, so there are those kind of language problems.

When you're talking about children's disability, you also run into these language problems with school records. School records are incredibly important for the disability determination for a child, yet, you know, you've got to speak -- you've got to speak special education, you've got to speak education in order to understand what -- what happens.

I had an example of this in -- in my own case, a case for a child where the child was being mainstreamed into one class. Previously to this, the child had been in a separate -- substantially separate class, small classroom,

with a lot of assistants, and he was being mainstreamed. And -- and the adjudicator was incredibly interested in this, thinking it meant that the child was probably no longer disabled; although, the adjudicator agreed that the child had been disabled before.

I don't speak special education, either. So I went back to the teacher, and she said, let me be clear. He's mainstreamed in science, he has a one-to-one aide. The aide tells him what's going to happen before he goes into the classroom. He goes into the classroom with the aide, and then she goes out -- she takes him out of the classroom and tells him what happened.

So he's -- you know, he was mainstreamed, but not really in the sense of a same age child without a disability.

So that's just one example of these kinds of language problems that -- that we bump into in these cases.

Also, functional capacity isn't really sought, especially not early on. In my

experience, medical information is sought, but treating sources and other professional sources are really not asked to provide functional information. What can the individual do on a sustained basis, day in and day out. What can the child do or not do that same age children without disabilities are expected to do. That information is not always sought. And that's another thing that advocates do when they get a case, actively seek that out.

Another problem that you really don't have any control over, unfortunately, is the difficulty of getting medical information out of sources or -- or information from any sources, really. A -- a prior panel member said that it can take months to get hospital records. And that's really true. Hospitals are, you know, trying desperately to keep nurses on the floor, and they do not lushly staff the medical records department.

Plus, you've got this overlay of HIPAA, I call it "HIPAA hysteria," where, you

know, scrutiny of your medical release is -- is common; you know, there's a lot of turnover in these jobs. So you get the partially trained person who is afraid her -- her hospital will be sued if she lets you have these documents, because it doesn't look quite right to him or her.

So there's just a lot of difficulty in getting medical information, especially from -- from -- from specialists, it's particularly difficult. Often the medical evidence that is kept in the ordinary course for the providers doesn't really answer the -- your questions, the questions you need to know, the questions I need to know to evaluate a case and decide whether to even take it.

And I have to go back to them for more information and -- and often it -- it takes a very long time to -- to get the definitive piece of evidence and -- and present it, and sometimes that comes rather late in -- in the process, which is too bad for the claimant.

I know that most of the DDSs have

medical relations staff, and they do spend as much time as -- as they can to be out there with medical providers and tell them about the importance of responding to the requests and doing it timely.

I think there's also some financial rewards if you do it timely, but apparently, it's not enough to get them all to respond on time.

I think medical expertise is often lacking to your adjudicators in -- in the disability determination process. You know, there's -- there's in-house doctors at -- at the DDSs, there are consultative examinations, people are contracted to provide these, and then there are medical experts at hearings.

And it's my understanding that the reimbursement available to attract these people probably isn't sufficient to -- to get enough or to get all of the right expertise.

You know, we've seen -- seen a lot of problems at the -- at the earlier levels, especially with -- with children, some of the rare

conditions that other panelists have talked about, nobody's -- the people haven't heard of them, they're -- the pediatrician doesn't enough to pursue -- provide the information that -- that Social Security needs. And, you know, the in-house doctors aren't aware of it.

I've seen children with, you know, really severe metabolic disorders, you know, get -- get denied when it was fairly obvious that -- that they should have been allowed if -- if the right information had been provided to the DDS examiner.

Often the DDS examiner has wanted to allow it but felt that, you know, they didn't have the medical expertise necessary.

You know, I've seen these problems over the years with all kinds of conditions which are now maybe not considered so rare. When I first started doing this 27 years ago, Lupus was a problem. People hadn't heard of Lupus, and I had to drag in people from the Lupus Society to my hearings to explain what Lupus was and -- and that

my client's symptoms and limits were typical of -- of people with that kind of a condition.

I've seen it with chronic fatigue, with HIV and AIDS, with reflex sympathetic dystrophy and -- and lots of others.

Sometimes disability evaluation rules that are helpful to the case are -- are -- are not follow -- followed, you know, the way -- how you weigh different medical opinions are specifically laid out in the rules.

Evaluation of pain and other subjective systems is one that's frequently misapplied. And the evaluation of mental impairments of all -- all -- all types of severity, really.

The last time the mental impairment listings were approved, I -- I mean were -- were adjusted, they were really approved, especially the -- the information in the preface where, you know, the -- there are some explanation about how Social Security looks at mental impairment disabilities and the kinds of tests that are

available.

That prefatory information is -- is really, really very helpful. It's helpful to me as an advocate. Sometimes I wish others would read it in more depth.

I also wanted to note that some of the Social Security rulings that have been issued in the last few years have been very helpful. There have been a number of rulings issued on conditions that have been difficult to -- to adjudicate. Chronic fatigue syndrome is one, interstitial cystitis is another, post-polio syndrome and reflex sympathetic dystrophy.

These rulings are really very helpful, in part because they explain the condition, they talk about what you need to make a diagnosis, and then what you need to determine the functional impact.

Now, it might be impractical to come up with a ruling for every rare condition, but perhaps a roadmap could be laid out that would help guide adjudicators generally for -- for rare

conditions, how you get the information and -- and then what you do with it.

One of the big problems that you run into in cases is what you need to determine that -- that the person, in fact, has a diagnosis or a medically determinable impairment, and then what you can use to evaluate the functional impact of that. It's very different, and often those rules aren't correctly done.

Existing expediting processes are -- are important. I think there's been some confusion about when what can be used. I think the QDD has worked wonderfully in Massachusetts, it's really great. We're happy to see that expanded.

I know our DDS examiners like it because they really feel like they can get benefits to people who really need them very quickly. On the other side, it's important to make sure that payment is also expedited; sometimes that doesn't happen.

But I think there's been some

confusion about whether QDD has supplanted other kinds of expediting, like the TERI process for terminal illness and presumptive disability, which is useful for people who are eligible for it, because they can get benefits; while -- while Social Security dots all the Is and crosses all the Ts for a formal determination, they can get benefits more quickly.

That's only available in SS -- initial SSI disability -- determinations, but that is something that people can really benefit from.

So I think that it's important to make sure all evidence is -- is collected from -- from all sources that have something to say about the impact of a medically determinable impairment on someone's condition, to make sure that applicants have help filing the paperwork. Many can't do this on their own and -- and don't do a very good job of completing the information without help; make sure applicants know what is needed to determine disability under the standard; try to provide more information to doctors, perhaps

working through some of these -- especially the -- the rare disease groups that are here today, to make sure they understood the standard and what they need to provide; collect functional capacity information much earlier; approve medical expertise in all -- all areas of the system; and perhaps consider providing more -- more guidance to your adjudicators in some of the important roles and -- and how to evaluate especially rare cases.

I find that particularly in childhood disability cases, not all adjudicators are -- are familiar with the standard in the evaluation rules. It's a relatively new standard, it's changed a few times over the years. And I think it's difficult to understand what the -- what the evaluation for -- for a child is.

I think most of us can understand trying to figure out what might prevent an adult from working, but it's hard to think about, you know, when a child has a marked and severe functional limit below what you would expect of --

of the same age child without a disability, it's just more conceptually difficult.

And I would recommend, as I said, providing more guidance through Social Security rulings, they've been very helpful over the years, and make sure that -- that the current expediting processes are -- are -- are clarified and people know how to use them.

I think it would be useful to expand the listings. It's always been my opinion that if something isn't in the listings, that is, sometimes there's a bit of a sense that it isn't as important or -- or not as likely to -- to be disabling. And certainly, having -- having more access to information about specific rare conditions and -- and diseases would be very helpful to -- to Social Security adjudicators.

Although I think you have to take care -- any time you come up with a list, somebody's going to take it as -- as the definitive list. So it's important that, as you expand lists or create new ones, to make sure that

all adjudicators understand that they're -- it's not all inclusive and that it's not intended to denigrate any other conditions that -- that aren't on the list.

Thank you.

COMMISSIONER ASTRUE: Thank you.

We're going to continue the policy of holding off our questions till the end. Although I do want to add one other process point, because your testimony reminded me. You veered close a couple times to subjects where we have ongoing -- we're taking comment on ongoing rulemaking, which is fine, but I just wanted to make sure that you and other witnesses know it's okay for us to actually talk about this, but we have a memo that we follow from the totally unreasonable general counsel of HHS that says we have to put those on the record.

So if -- when we go into the Q and A, I don't want to inhibit any conversation, but I feel honor bound to let you and others know that if we actually do cross over the line as something

that we're taking comment on now, our lawyers will have to sort of summarize that and put that in the official administrative record.

So I just wanted to --

MS. LANDRY: Don't want to make more work for the lawyers.

COMMISSIONER ASTRUE: Yeah. I try to -- try to make it as easy for them as possible.

Okay, thanks a lot.

MS. LANDRY: Thank you.

COMMISSIONER ASTRUE: We'll move on now to Mr. Leach.

MR. LEACH: Good morning. My name is Bill Leach with the A.C.C.E.S.S. Program in Tampa, Florida.

I want to thank you for this opportunity to talk to you today about issues from disability determinations for people with rare diseases.

Excuse me. Since 1995, I have represented hundreds of claimants with rare chronic conditions before the Social Security

Administration.

Our program provides representation at no cost to these individuals as -- that are part of certain specific disease communities.

My comments and suggestions are based on this experience and are focused primarily on the disorders in which our program specializes. I should emphasize that both Kim Bernstein and I are speaking here today on -- in our capacity as advocates for the disabled and not on behalf of the company that funds our program.

I would begin by talking a little bit about the -- the -- the TERI cases. My experience with those is that they generally don't need me. If it's a TERI case, I can pretty much tell them what they need to do and have them get in touch with the local office, and I don't hear from them, because basically that's a success story.

So, you know, the -- the -- the TERI process where it's working is working well. Most of my clients don't qualify for TERI or for some of the other like presumptive disability cases,

and so that's mainly what I'm going to be talking about today.

It's not that I don't get those cases, it's just that I have very little interaction with those clients because, for the most part, they -- they get what they're looking for.

In my experience, people with rare diseases who unquestionably qualify under the listing of impairments can have widely divergent outcomes. One of the things that I often tell our clients is that I can take the same facts and put them in front of two adjudicators and get two different results.

Some claims are approved quickly, some take longer than anticipated, and there's really no reliable way to predict how long your claim is going to take. Moreover, the lack of predictability in the actual outcomes leads to a perception in the general population that the process isn't always fair.

Part of the disparity arises from the fact that some of the listings are open to

interpretation. There's one that's in the process of being revised, and I should probably be careful about talking about this, based on what we just said, but there's one for what's referred to in the adult listing as a cell mediated immune deficiency which talks about documented recurrent severe infections. And there is an ongoing process to revise that into something a little more specific.

The problem is that there's never been a definition of severe. And so in hearings, we often get into a question of what is or isn't a severe infection for somebody that has no immune system.

And this is something that reasonable minds can differ on. And so this is one of the reasons why we get disparate outcomes is that the listings themselves are often largely open to interpretation where there's a lot of terms used that aren't really defined.

And, again, the process of going through the listings and trying to sharpen those

and clarify those, I think will help to minimize some of those issues as -- as we go forward.

One of the problems I've been hearing a lot about lately is determining which listing has been met and at what time.

We've talked a little bit about amyotrophic lateral sclerosis, ALS, or Lou Gehrig's disease. I've had a few calls recently from people who were approved for disability for a diagnosis other than ALS and that were subsequently diagnosed. And this is not uncommon. People with ALS have severe health problems, long time -- long before they're actually finally diagnosed.

The problem is that they're being told now that they have to go through the 24-month waiting period for Medicare because they were not originally diagnosed with ALS as part of the determination for their disability.

Since we're talking about compassionate allowances, one of the things I would like to suggest we maybe think about is

using that as the process to amend favorable determinations to include a diagnosis where we can medically relate it back, showing that the ALS was in existence at the time the determination was made. Right now there's no good way to do that. We're just sort of doing it on a catch-as-catch-can basis.

This would allow people with ALS to get into the Medicare system sooner than the 24 months that is typically required, and that's because typically, they don't survive 24 months. Now, typically by the time they qualify under the -- the old law, the Medicare card came too late for them.

In some instances, the delays in allowing claims for people with rare chronic conditions arises from what's been referred to as the translation problem.

I've also often talked about it as -- as a -- basically a translating from one language to another, basically from medical to regulatory.

Part of the problem, you know, we --

we've talked about the fact that Social Security doesn't speak in medical terms, and certainly that's -- that's at least one part of the problem.

The other part of the problem is that doctors don't understand the disability process. And I think that Social Security can maybe do a little bit more about reaching out to them and getting them to understand the process that Social Security actually uses to determine things.

It -- it's sort of an interesting puzzle where at the one end I've got a DDS adjudicator who's trying to read handwritten progress notes from the doctor, trying to figure out what the doctor is talking about, and at the other end, the doctor is scratching his head, looking at an RSC form, saying how could do I fit my -- my patient into this form?

So I think that there needs to be more of a -- an outreach and -- and cross-communication between the medical community and the Social Security Administration to talk about these functional limitations that we've been discussing

all day, because that's one of the biggest issues in terms of not only getting the medical evidence in in a timely fashion, but getting it in in a form that Social Security can use it.

And I also want to tag on to what's been said earlier about the folks at the DDS. These folks are doing the best job they can with what they got to work with. Anything that we can do to make it easier for them, I'm all in favor of. And I think part of that would be to help them understand that there are conditions out there that they're only going to see once or twice in their entire career, as opposed to the bad backs that they see every day, and that there needs to be special resources available for -- to help them adjudicate those types of cases.

One of the examples that I gave in my written testimony about this problem of translating, there's a listing for what I refer to as pulmonary hypertension, it's listing 3.09A. The listing actually calls it cor pulmonale secondary to pulmonary vascular hypertension.

A lot of people that meet this listing get turned down, and the reason why I think is because a lot of these buzz words do not appear in medical records.

I rarely see cor pulmonale showing up in pulmonary hypertension medical records. And it's interesting because in the explanatory material that's in the -- in the front of that section actually talks about the term that's more commonly used, which is right ventricular hytropathy -- hypertrophy, sorry, and -- but it's not in the listing.

And the other problem is that the condition, as I said, is typically referred to as pulmonary hypertension, not pulmonary vascular hypertension.

One of my suggestions is that if we could create a database where adjudicators had access to the ICD-9 codes that are used for insurance purposes, medical purposes to identify medical conditions, this would help them to identify cases where it requires special

consideration where there's a particular listing that they may not be aware of that would allow them to quickly and easily pull up perhaps a specific questionnaire designed by medical professionals familiar with that particular condition that can then be sent to the treating physician, so that when we request the medical records, we not only get the medical records back, but we also get a questionnaire that does fit the patient when the doctor looks at it.

And that's something that I think can -- can be implemented simply by requiring that we move from the Fussell (phonetic) codes to the ICD-9 codes so that we've got a consistent system for identifying these different disease states.

Taking pulmonary hypertension as an example, the questionnaire could be used to identify claimants who use medications that require continuous infusion pump through an implanted catheter, which is a -- a common therapy for that particular condition. Or they require frequent daily use of a very specialized

inhalation device that, again, has a lot of different requirements.

Or, I mean, some of the other criteria that we're looking for under TERI and so forth, such as a need for oxygen to function, suffer from chronic pulmonary or heart failure or have been placed on a heart lung transplant list. If -- if we have some way of quickly and easily identifying these cases, it would make it easier for adjudicators to know, okay, there's a special process for claims with these type of facts in them.

Finally, one of the problems, and I -- I hesitate to say this, but I really don't know any other way to say it, there are some listings that the adjudicators just don't like. The -- the listing that I have the most problem with is for children with inherited coagulation disorders, hemophilia, von Willebrand's disease, this is listing 107.08A. And it's fairly simple. It provides for disability where there is repeated inappropriate or spontaneous bleeding. And that's

all it says.

I just represented a child in New York where the medical expert testifying at the hearing said that she didn't meet the listing, the child in question, despite the fact that she admitted my client had repeated bleeding episodes, but she hadn't been hospitalized recently. And I said, well, Doctor, there's nothing in the listing that says that, and she says, I know, but she doesn't meet the listing.

So some of the listings are just not favored. And other than telling people we really mean what this says, I don't know what else can be done about that problem.

One of the issues specifically with people with coagulation defects, both adults and children, is that they frequently treat at home rather than a hospital or clinical setting.

Treating physicians routinely rely on these home treatment records to prescribe for their patients. Allowing adjudicators to rely on home treatment records is -- just as the informed

medical professionals do, would help expedite favorable outcomes for people who are often approved on appeal.

In denying a claim, adjudicators often talk about the fact that the claimant is under -- undergoing some form of treatment that lessens the impact of their disease; however, they hardly ever address the extent to which the relief is provided by this therapy. Very often, the treatment takes a horribly debilitating condition and makes it slightly less horrible is by no means a cure, and their benefits are usually offset to some degree by the impact of the therapy itself.

What I would suggest is that things like the frequency, duration and side effects of these treatments are rarely considered in the evaluation of a claim as functional capacity, until typically we get to the -- the ALJ level in the hearing process.

As an example, it's very difficult to maintain regular work attendance when you're going to require a six-hour infusion every three weeks

for the rest of your life and the treatment leaves you feeling ill for a day or so. This is a common pattern for, again, people with primary immune deficiency who are using intravenous immunoglobulin to treat their condition.

Another group is similar pattern of infusion is people with a genetic form of emphysema called alpha-1 antitrypsin deficiency.

The other thing that we often don't talk about in terms of functional limitations and treatment is how long does it take for the treatment to work. People with coagulation defects can suffer from extreme pain and restricted range of motion in the affected area for days before a bleeding episode is finally controlled. The sporadic and unpredictable occurrence of such episodes has a huge impact on their ability to function.

These people with bleeding disorders such as hemophilia and von Willebrand's disease should be considered for compassionate allowance when the records demonstrate spontaneous bleeding

episodes that require treatment, and I want to suggest six times or more a year, because the functional impact of that is such that they would not be able to maintain regular employment or to maintain age appropriate functioning in -- in the case of children.

I do think that the Social Security Administration needs to continue working with medical professionals who specialize in rare disorders to improve the listings. Again, I'm talking about hemophilia, which is probably about half of what I do. Adult listing 7.08 for coagulation defects looks for transfusions of whole blood, despite the fact that the prevailing treatment for this condition today consists of infusions of blood clotting factor. As a result, the listing is rarely met on -- in -- on terms of the language that's now being used.

From a process standpoint, my feeling is that the pre-effectuation review by the regional offices of half of all SSDI allowances discourages complete claim development,

particularly since the odds of having a denial reviewed are slight.

I question the value of these reviews, and I would recommend substantially reducing, if not eliminating, them outright.

On the flip side, we all know that it costs more to approve a claim after an appeal to ODAR then to approve it on an initial application, but that added cost is not borne by the responsible DDS. There needs to be some sort of disincentive that makes the denial of claims that are eventually paid as dreaded as the pre-effectuation review.

This ought to include some sort of feedback to the adjudicator that denied the subsequently approved claim.

I also want to give credit where credit is due. Many of the initiatives that the Social Security Administration has undertaken as part of the Disability Service Improvement plan should help speed up the process.

I do sometimes have some -- some

concerns about that, because a quick decision is not always a good -- a good decision.

I just talked with a client the other day about the fact that she was wanting them to hurry up and -- and get her reconsideration done, and I said, listen, we're better off getting a good reconsideration now then waiting two years to get a favorable decision from the ALJ.

So speed is good, but we also still want process -- you know, quality process to take place as part of the review.

The big problem that I think has been alluded to has been the lack of adequate funding for the Social Security Administration and for the DDSs. The hearing capacity is woefully inadequate for the number of claims that are pending for hearing. We've now got cases waiting for hearings two years and longer in many parts of the country.

I realize that this is a legislative problem rather than a regulatory one, but it's probably the number one reason why so many people are waiting too long to be paid benefits that

they're actually entitled to.

Thank you very much for this opportunity to speak with you.

COMMISSIONER ASTRUE: Thank you.

MS. BERNSTEIN: My name is Kim Bernstein. I would also like to thank you for the opportunity to speak. I'd also like to thank you for all hard work you're doing.

I -- I originally became involved by listening to a presentation that Ms. Braunstein did at the NORD conference.

I am the director of the program and have been for the past 13 years. Our program represents people, as Bill said, regardless of choice of provider and free of charge. Unlike Bill, I stopped doing hearings quite some time ago.

I would say I'm in the recovery from the practice of law. Instead, I do the advocacy work. And I've been speaking around the country, basically every other weekend, for the last 13 years for many, many groups of people who need

disability benefits.

I think that we've spoken a bit about the convergence of translating medical information and regulatory information in order to get benefits, but the third thing I think we're missing is the language of denial, because what we find so often is that with these conditions, it really isn't a matter of is somebody faking it, although they may feel that; it's a matter of have they met the level of severity, do they have the medical records, and when they get to the hearing, are they a credible historian of their condition?

My biggest concern is in -- in the psychology of denial, we have people with can't do bodies who have can do attitudes. And it takes an awfully long time just to get to the point that they say that they're disabled or that they can say that at a hearing.

I actually had somebody at a hearing who is a quarry worker with cystic fibrosis who, when asked, said, yeah, I could probably work. His pulmonary function studies show he probably

couldn't.

But that's one of the problems that we have coming in, is that people who almost can't say I'm disabled, because disabled brings up broken, dead, half dead or inability to have a good day.

I also find a lot of mothers, and I do think, just like the words for snow in -- in the Eskimo language, there should be a different word for mother when we look at the mothers of the children with MPS and other disorders like that. The mothers who often say I don't want my child to be disabled, so I'm not going to file for benefits until they've pretty much bankrupted the family.

But what I'm concerned about is that the concerns that I've heard are, first of all, people with rare chronic conditions find so many ways of coping that when they say I can do laundry, I can go to the grocery store, often it doesn't really mean that.

I've had somebody tell me they could go to the grocery store and do their ADLs, when it

turns out what they did was they went to the grocery store with their spouse, sat in the front of the store while the spouse shopped. They say they can bring the groceries in; really, they sat on the front step.

So the information that you're getting to start with isn't really the real information of what their bodies can do, it's what their minds can do. Because for them, they're still grocery shopping.

And I think it's very important that our forms really ask the questions in ways that are broken down and that we ask the questions rather than just waiting for people to answer them. And I think that's part of what Bill was talking about.

People say I can do the laundry, but we don't ask how long it takes, and I think we have to have some questions that will sort for denial.

Somebody with multiple -- with myasthenia gravis once told me that means he

doesn't even know it's a lie, and I think that's a big -- big part of the problem that we run into.

I think that when you have the initial claim denied, it feeds into the feeling of denial, especially for people who had faced a long diagnosis process where the claim is denied and they say, you know, I'm really not that bad.

I had a friend who had hemophilia, HIV, HCV, several joints that were fused and others that were replaced, tell me that he thought he could probably work and wasn't really sure that he was entitled to benefits. And I just think we have to understand that going into the process.

Many join the large number of applicants who don't appeal the denial of their claim because not only do they have to focus on the reluctance to say I can't, but the feeling that they may not be entitled. And this causes them to -- to stop and then start again later and go through the process.

I also have grave concerns about women with rare chronic conditions because, again, they

also have a long period of diagnosis. Often they're told it's in their head until the diagnosis is finally found, and then when they don't work for five years, after the first year, they're involved with children, by the time the fifth year comes along, they lose their insured status and their excuse to say that they're not disabled, and then they're outside of the time when they'll actually be able to get benefits.

I'm also concerned that there's a real need to expedite determinations and fully consider functional limitations at the earliest part of the process for those with rare chronic conditions, partially because the -- the DDSs don't understand these -- these conditions.

Half the doctors don't even understand them. So we need to find a way of looking at the functional capacity at the earliest possible step rather than at the administrative law judge step where the administrative law judge will find a way of understanding the residual functional capacity, but so much later.

I think that if we can do more with the RFC at the earlier stages, we'll probably have, as Bill said, a savings and an offset.

I'm convinced there are solutions, and I also believe that the savings realized from avoiding unnecessary appeals would at least partially offset the costs of implementing new procedures.

I would also propose transitioning the use of ICD-9 diagnostic codes, which would make it easier to identify the rare conditions that could qualify for compassionate allowances. Because it will take a long time to implement, I would ask you to consider expanding and improving the current methods for flagging the rare chronic conditions.

I would ask you to work closely with the medical professionals who specialize in rare chronic conditions, but I'm also concerned that a drop box may not be enough, that it may take human contact and words in order to explain this to the -- the people making the decisions at the

lower appeals.

This can -- if we -- if we use the questionnaires that are designed for more appropriate treating rather than the one-size-fits-all, I think that we will accomplish that a lot sooner.

I think we need to -- and -- and this is, I think, the thing that I'm most -- I'm most convinced would make a difference. When we can't adjust the wind, we've got to adjust the sails. And I don't think that we can overhaul the entire Social Security Administration's processing, but I think we can meet it with better advocacy and advocacy that's available to people with rare chronic conditions.

Our programs are rather unique in that we only deal with rare chronic conditions. We understand Social Security; we understand the disease; we understand the people.

And I think we need to have more groups available to people with rare chronic conditions who understand all three to assist the

Social Security Administration in making decisions well and quickly.

If we were to establish a panel of consumer advocates for rare disorders, either within Social Security -- I did find when I was a public defender one good prosecutor was better than 100 good public defenders, because if you have the person making the decision more educated and more able to handle it, the decisions come out better and faster, and it transfers to everyone else -- or from an agency such as NIH, Office of Rare Diseases or from an independent outside group, for example, National Organization of Rare Diseases, if we could house a group of specialized advocates that could assist other lawyers by providing briefs, as we often do, to people who have hemophilia and have not asked for our services, they could assist in identifying the functional limitations, developing compassionate allowance guidelines for disability claimants with rare diseases.

These consumer advocates could also

make sure that unrepresented -- and they concern me more than anyone, is the unrepresented client with a rare chronic condition that hasn't yet found the nonprofit organization which will explain things to them and has not found anyone to help understand that what they have will be understood if they keep going.

We need to find ways of offering people that will help perhaps in a pilot project that would assist the -- the Agency as well as the clients who are applying.

Every denial letter needs to include a disclosure of what percentage of claims on average are approved or denied at each stage of the appeal process and what the average processing time is for -- for the appeal.

I think people get the letter and they say, see, I wasn't really entitled. I was wrong. Especially after they've spent so much time getting a diagnosis where they were told, get a new job, something, it's you, it's really not your condition.

It should also make clear that -- what the relevant waiting periods are for cash benefits and associated medical coverage from the date of onset. My feeling is that every month of delay in approving a disability claim is ultimately -- that's ultimately allowed should result in corresponding reduction in those waiting periods. We've got to find a way of getting the benefits to the people.

We are so grateful to have the invitation to come speak. I really believe that the main thing is, we can't give up. We need to find creative and collaborative ways of bringing the stakeholders together in order to get a better health outcome.

I think we all share the common goal. If we can join advocates, attorneys and nonadvocates, the medical community, the voluntary health organizations, the governmental agencies, and I think something novel, corporate partners -- there are so many corporations that put so much work into finding therapies that serve the people

to give them better lives that need reimbursement sources so that the people can actually utilize them. I think that we could take some dollars from the corporate side, bring it in and find a way.

Once we have a diagnosis and an explanation within the time benefits, within the time frames, people will get their benefits and will get their products and enjoy a much healthier life.

I think we also need a lot better lookback at last date insured, especially for women. That's something that very much concerns me. I didn't quite understand it until I was a mom, how the first year you could say, I can't be disabled. I don't care what you call me; you cannot call me disabled. I have a child to take care of.

And by the time you finally realize the child is not at home and you weren't doing what you really needed to, you were soliciting help from your family, your relatives, you most

likely won't get your benefits, and that doesn't help the family. Those are benefits that you're entitled to.

We need better lookback. I think it's probably going to take putting private dollars in for representation, but I think they're there.

I want to thank you again for giving me this chance to speak. If you ever decide to consider changing the name of disability, I think it would be a great thing. I think that really often we're looking at inconsistent worker status, and I think that the people who could get inconsistent worker status would be much more likely to apply, work when they could, and not when they couldn't.

Thank you so much for giving me this chance to speak.

COMMISSIONER ASTRUE: Thank you very much.

I wonder if I could ask the panelists to -- to help me with a problem that I'm still trying to get my arms around, because I've -- I've

probably had a typical experience, at least with physicians that help patients who rare diseases.

I've always found them to be extraordinarily zealous advocates. When -- when I was in biotech, I had the experience of being cornered in parking lots by doctors that were trying to demand that their patients get into clinical trials and that type of thing.

And so the -- the -- the question of getting medical records, I -- I guess I understand a little bit more in some of the larger general practice facilities.

I wonder if you could all give me a little bit more of a sense for -- for this particular population that we're focused on today, the rare diseases, is it a real issue getting medical records on a timely basis? Does it depend on specialty or the things you could do?

If there's any way you could kind of focus on that issue specifically for this population, I'd be grateful.

MS. LANDRY: I -- I think that it

varies. I mean, certainly I've had contact with -- with specialists, children's doctors in particular, just had a case involving Rhett's syndrome where the doctors were very responsive, gave me their cell phone numbers.

But could I get a letter out of them? Could I actually get them to put pen to paper and an -- answer the questions that -- that I had asked? That was harder.

And I think part of it is this language issue. They would write me a letter, but it really wouldn't respond to the questions that I asked. They wanted to say the same things over again that they say to their college -- colleagues and that they put in -- in -- put in their already-prepared -- the preexisting medical records.

It was hard to get them to answer the questions around functional capacity for the child or the specific medical questions. They wanted to, they tried to, but it -- it took a while for me to get that information from them, despite

their -- their cooperation.

And -- and sometimes, you know, doctors are only willing to give you the existing medical records. They're not willing, without charging you, to -- to provide a -- a -- a statement. I mean, I'm a nonprofit. I can't -- I can't pay 2-, $300 for a prepared written statement or -- or an RFC.

So, yeah, it -- it really varies. A lot of doctors are really helpful, some doctors write more than others. I find the higher degree of specialty, the less they write. You know, the -- the therapists will -- will write, you know, the psychologists will write, but -- but others, it's -- it's harder.

So, you know, over the years what I've tried to do, and I'm sure you have, too, Bill, is to come up with questionnaires and check sheets that are very, very specific, and send people a copy of the listings, you know --

COMMISSIONER ASTRUE: That works -- does that work substantially better, in your

opinion, if it's a more automatic process like that?

MS. LANDRY: Well, I mean, it does to the extent that the information that you need can be had that way. When -- when what you need is -- is a little harder to get at, a little less objective, perhaps, if it's functional capacity information or some of the softer information in -- in the -- in the listings, it's, you know, like severe, what does severe mean? Everybody has their own definition of what severe means or what disability means, for that -- for that matter, what sedentary means.

And so you -- you spend a lot of time explaining what Social Security means by these terms, and it's -- it's just hard to get them to -- to respond and to -- to what you want. I mean, moderate means something different to them then it does to Social Security. A moderate level of osteoporosis is pretty darn severe. It doesn't mean, you know, sort of severe. So you spend a lot of time explaining language.

MS. BERNSTEIN: I think there's also a problem in positive charting, where people are trying to chart positively in order to have the medication and have their amenities. Or, for instance, for someone like Ralph, who has three fused joints, if he's walking, he's ambulating well. So that's what goes into the file to Social Security, not really a good picture.

MR. LEACH: Yeah. And just to -- to add on to what's been said already, I -- I find that it really varies a lot in terms of getting medical information in a timely manner. Part of that has to do with the fact that, yes, we can pay for records, but we have a very convoluted system for doing it. And it involves sending tax forms back and forth and getting them to sign the forms so that we can issue a check, and then waiting for that check to clear, and then waiting for the medical records people to pull the files off, and the medical records are handled by an outside agency that's going to do it on a first-come, first-serve basis.

I -- I spend months trying to get medical records out of sources very often. Some doctors are really good about it, they'll fax their medical records right out -- over; others send me a bill and say, you know, we'll send you the records when we get your payment.

In terms of trying to get the specific information --

COMMISSIONER ASTRUE: Is -- is that legal? You -- do you operate just in Florida or do you operate in other States as well?

MR. LEACH: We -- we represent people all over the country.

COMMISSIONER ASTRUE: All over the country. I mean, in most States, I mean, I think if -- if a patient requests medical records, isn't the physician required to supply it?

MS. LANDRY: They can charge.

COMMISSIONER ASTRUE: They can charge them?

MR. LEACH: They -- they can and they do, believe me.

COMMISSIONER ASTRUE: And what's -- and what typically is the charge?

MR. LEACH: It's --

MS. BERNSTEIN: Varies.

MR. LEACH: -- it varies a lot. Hospitals tend to be very high. They'll want to do a per page charge, and hospital records tend to be voluminous. I just paid -- or not me, personally, but our program just recently paid something upwards of $300 for medical records for a child.

So it can be very expensive, particularly when we're talking about people that don't have the financial means to -- to -- to do this on their own. Like I said, we -- we have a -- a corporate sponsor that pays for this stuff, but we have to go through their channels to get the checks issued.

As far as getting the specific information that we're talking about, really, these questionnaires and so forth --

COMMISSIONER ASTRUE: Well, why don't

we hold and let the bell ring. I think -- since people are trying to listen long distance. I -- I know from experience, it doesn't last long.

Yeah, we have a historic church right next door.

Okay. I think that's faded, so --

MR. LEACH: Okay. As I was saying then, in terms of getting specific questions answered, I -- I have to tell it as a war story. There's a lady that I represented on pulmonary hypertension who met the listing, but we didn't have the language "cor pulmonale" anywhere in her medical records. And I kept trying to get the doctor to write me a letter that says she has cor pulmonale.

And she went through all these convoluted -- we tried to -- and she -- she ended up losing her insurance, moving in with her adult son because she couldn't keep the mortgage up anymore.

The day of the hearing, I -- like three days before I got the letter from the doctor

that said cor pulmonale, the judge looked at the letter and says, oh, it says cor pulmonale, we don't need a hearing now. Because that was the only thing that was missing as far as the judge was concerned is that we didn't have that specific language in the medical records previously.

So, you know, we -- we're kind of at the mercy of medical providers in terms of getting records in terms of their financial requirements, their legal disclosure requirements, because HIPAA's made it much more difficult.

For every medical provider out there, there's, you know, an independent interpretation of what a -- a medical release has to have in it, and we spend a lot of time sending medical releases back and forth saying, well, will you accept this? Will you accept this? Why don't you send me one of yours and I'll have him sign your release and then send it back to you. And all this stuff takes time.

COMMISSIONER ASTRUE: Okay.

MS. LANDRY: Absolutely.

COMMISSIONER ASTRUE: David, you got anything?

ACTING DEPUTY COMMISSIONER RUST: Ms. Landry, you made a passing reference to the idea of expanding the -- the listings.

Could you give us a little bit more detail and follow-up on that a little bit?

MS. LANDRY: Well, to the extent that some of these rare diseases and conditions that probably clearly meet the listing would -- would meet listings level severity aren't in the listings, it would certainly speed up matters if they were. That's -- that's really what I meant.

I mean, if -- if you can actually get the medical criteria for a listed impairment -- and -- and often you can -- I mean, we don't see too many listings cases that come out of cases that should have been listing allowances that come out of our DDS, so I got to think our DDS catches a lot of them. They're able to get the specific medical criteria that you all agree meet your definition.

And so to that extent, it would be helpful to have more listings for these conditions.

MR. LEACH: One specific suggestion I have on that, and this was something that came up before at a policy conference for the immune deficiency listings.

With primary immune deficiency or cell mediate -- mediated immune deficiency, there's roughly 100 different disorders that fall under that umbrella, there's not a list anywhere in the explanatory material that says which conditions are considered primary immune deficiencies. And even just a list saying no -- if -- if it's one of these conditions, hypogammaglobunemia, you need to look at primary immune deficiency as the listing that applies in that situation.

So even just a laundry list that says, you know, this may be the appropriate listing to look at this for this particular condition would go a long way towards streamlining the 7,000 number that we're looking at, because many of

these conditions overlap.

DR. GROFT: I -- I got a couple of questions, and I'll make some statements before I -- I ask you for some answers or just responses.

Are there groups of diseases or rare diseases that -- that particularly are not represented within the ICD-9? That would be one of the first questions.

And then, how frequently do you really run into this situation where they're not included?

And then just to give you some of the background information, I mean, I think all of us know how difficult it is for many of the rare disorders it is to really obtain the diagnosis because of the expression of the disease and that it affects multiple organs, and you always don't get those clearcut diagnostic criteria, boom, or even imaging results right away that says this -- this is this rare disease. So it is a particular problem.

But I do want just to mention, too,

that the World Health Organization has established an advisory group on rare diseases that we are looking at revisions in the ICD. Now, this, I think, will not be completed till around 2015 for the ICD-11. And I've got to think. I know I -- I'm well beyond the Social Security eligible -- eligibility at that time. But hopefully, we can keep working on it until then.

We can put -- but I think there are a number of us that want to do something that we can address this issue immediately, that perhaps we can get some information out more readily.

So if you have any thoughts at all about -- are there particular groups of disease that we should look at first off, that would be helpful. And then to what extent is it a problem.

MR. LEACH: It is a problem in that there are, as you said, a lot of conditions that don't have specific ICD-9 diagnostic codes, or a lot of times more than one code can apply, depending on how the medical evidence is interpreted and -- so it's hard for me to come up

with a specific one and say this -- this is one that -- that's not included.

I would say, in general, that anything that moves us toward helping adjudicators to identify rare chronic conditions and what part of the regulations might be applied to them is going to be beneficial.

We're not going to be able to do it perfectly, either now or in 20 -- 2015, but we can at least do a little bit better now than we're doing currently in terms of saying, you know, there is a specific ICD-9 code for this particular condition, and these are -- these specific functional limitations that experts tell us are often associated with that condition.

Like I said, I -- I think that there are, you know, a lot of conditions that I'd like to see listed. But the other -- you know, the other side of that, again, comes back to the same thing I was saying about adjudicators is often true of specialists. Sometimes they don't see more than a couple of patients with these

conditions as well, and so they may not be familiar that there is even an ICD-9 for that particular condition, or, you know -- that they may not see it enough to -- to know much more about it than what's in, you know, the medical literature about it.

So they -- they don't often have a lot of the -- the treating experience that would allow them to address it in the fashion that we're talking about, either.

That's one of the problems we run into with rare conditions is that there's not enough people with these conditions where we can really come up with a consistent approach, saying this is going to work in all situations for all people with rare chronic conditions because, A, they're very variable and, B, it's so unpredictable as to which person's going to have it.

MS. BERNSTEIN: I think as you go through those, it's very important, again, to include the -- the community of advocates. When you look at, just say, okay, this is what we're

saying from a medical standpoint and make sure that we're including the people who will take the listing and then try to turn it into benefits, as well as the voluntary health -- the voluntary health groups that understand the people and, again, the -- the denial that goes along with it, and what you're not going to see in the medical records and need to ask for, both of the person and of the physicians.

DR. GROFT: Thank you.

JUDGE CRISTAUDO: A number of questions about functional capacity and some -- and about gathering evidence --

COMMISSIONER ASTRUE: Little closer to the mike.

JUDGE CRISTAUDO: -- a number of questions and comments about functional capacity and about collecting evidence.

There was one comment, I believe, that suggested something like we need to be looking at functional capacity in the prehearing stages.

Ms. Landry, I think you may have said

something like that, or if I misunderstood that, I apologize.

MS. LANDRY: I -- I did. I -- I -- what I meant, and I'm sorry if I wasn't clear, was that, you know, more functional capacity, better functional capacity needs to be collected earlier on in the process at the initial and reconsideration levels, along the lines of what -- of what Bill testified to, ask more specific questions, you know, really go after it in a much more targeted way.

JUDGE CRISTAUDO: Is -- is there a better way to do it than the way we've been doing it? I mean, we ask for functional capacity information obviously -- obviously apparently more at the hearing stage than we do perhaps at the prehearing stage, but is there a different way to do it?

MS. LANDRY: Well, I think -- I think at the initial stages, large -- you're -- you're always starting with -- with a form. And so the forms could be better. The forms could ask more

information.

I think one of the areas that is almost never collected is the effect of fatigue or the effect of -- of treatment on people. I think Bill mentioned this, as well. You know, a lot of folks are -- are able to do something. I remember one judge, Federal Court judge said, you don't have to vegetate in a dark room to meet the Social Security disability standard.

So, you know, people are able to -- to do some things. And -- and the -- the trick is to figure out when they can't do enough to meet -- to meet the standards.

So often people are, you know, doing household chores. This comes up a -- comes up a lot, but how do you do them? In an -- and -- and the sense that it's -- there's no sense that it's different to be able to do household chores in your own home on your own time and take a break when you need to then to be able to be on the job, you know, with your supervisor breathing down your neck, seven hours a day, five days a week on a --

on a consistent basis without too many absences.

So that -- that kind of information isn't -- isn't collected. It typically isn't collected until, in my experience, you -- you get to a hearing, especially if you have -- you have an advocate who, you know, sort of looks at that and -- and has -- is -- understands your condition.

I think a lot of rare chronic conditions contain this -- in -- include this sort of feature of -- of -- of fatigue and the need to rest and -- and sort of very debilitating treatments.

COMMISSIONER ASTRUE: Let me just cut in for a second. I mean, we are -- we have been trying to look at that in -- in a very hard way. And -- and to the extent that we can find ways to make it objective and easy, we're trying to do that.

I mean, one of the nice things that the staff put into the recent digestive regs is that we collapse most severe forms of liver

disease and said if you have a diagnostic -- a common diagnosis -- diagnostic score for liver is called the MELD score, and it's a composite of three different elements. And if your MELD score is 22 or higher, we're not going to bother going through the individual analysis and with all the indeterminacy of that, and we're just going to say it's on the basis of the medical evidence, we can conclude conclusively that you can't stand and do the various things that you're required to do for functional analysis.

We're not going to be able to get away from collecting that information in -- in the vast majority of the cases. And there's always going to be issues with that. That's -- a lot of that is probably an unsolvable issue.

But we are looking on a disease category-by-disease category basis to see if there are other generalizations that we might be able to make, particularly -- this gets outside of rare diseases, but particularly for some of the -- the very large diseases where we see

recurring cases.

So one of the things -- you know, we've been talking to NIH about a couple of issues, but one of the ones that I'm very interested in is looking at imaging techniques that -- that -- that measure blood flow to the heart, because I do think for a wide range of cardiac diseases, it may be possible to use new technology to draw some lines that would make some of that anecdotal evidence irrelevant in particular cases.

Again, we can't do that for everyone, but we may be able to do that in a fairly substantial number of cases and greatly improve the process.

So this is a fairly new thing for us, but it's something we're very open to, and if you've got suggestions as to where we might be able to draw those lines for other categories of cases, I encourage you to let us know.

And we're also getting away from only doing the comprehensive reviews. So if we've got

two or three things that we need to clarify in a listings reg, you know, we're not going to wait for, you know, the -- the cycle of our recent history, which can be a very long one, to update them.

We're -- we're -- we're -- we're willing now -- we haven't done it yet, but we're in the process of getting started to do that, to do some relatively small adjustments in the regs where it makes sense for us to do it.

So if you've got recurring categories of cases where you think we might be able to cut through some of these things, we're very open to taking a look at that.

MS. LANDRY: That's great. And I -- I think it's very helpful to do it that way. I think it would also be helpful to note where these kinds of limits can occur with particular range of -- of diagnoses, even if you can't come up with the objective test, is to just let it be known that it is a consideration and, you know, they can do the usual evidence gathering and credibility

findings that you have to do when it's not an objective test.

MR. LEACH: Responding back to the -- the -- the judge's initial question about the -- how can we improve the RFC evaluation, the biggest issue that I see with the forms that are sent out is that it asks, you know, how -- how long can you walk? how long can you stand? how long can you sit? lift? carry -- various activities. It never asks, can you do this all the time or are there times when you can't do this?

And I think that's the biggest issue, is that many of our patients with rare chronic conditions focus on when they can do something. Yes, when I'm feeling good, I can walk a block. Let's not talk about those two or three days out of the month when I stay in bed all day because I can't do that.

So I think it really needs to be focusing more on the good days/bad days type of thing, because I will tell you that once it gets to the hearing level, that's typically where

the -- you know, the judges want to go, is talking about how often can you do these things and what are the limitations there. If we got that information into the system earlier, I think it would improve the adjudication process at the lower levels.

MS. BERNSTEIN: It's also a matter of quality, quantity and duration, because if you -- one of the first cases that really made an impression on me, if the spouse hadn't been there, the gentleman said that he could cook for himself; he could shower by himself; he could go grocery shopping; he could go ice fishing; and he made peanut butter and jelly. He -- he couldn't do anything.

But nobody asked him until we got to the hearing, what do you cook? How long do you stand? When you shower, how long does it take you to recover? And after ice fishing, how many days are you down?

And that's -- I mean, that's the main thing I find, it's gardening or it's yard work,

and how many days does it take afterwards.

But I'm sure you ask those questions at hearings. I have always seen that. I've never seen an unrepresented hearing, but I'm sure that those are the questions that are asked when there isn't an advocate, and those are the questions that need to be asked when there isn't an advocate at the lower levels.

COMMISSIONER ASTRUE: Yeah. I -- I -- I've -- I've sat in on -- on some hearings recently, and -- and at least the ones I've seen, they tend to do a pretty good job on that. And it is -- at the end of the day, it's an art rather than a science. There's -- there's judgment that we ask the judges to use, and we can't break it down, so it's -- because every -- at some point, most of these cases are very different, and you've got to just rely on your experience and your intuition to get the right kind of information out.

And it is sometimes very surprising, you know, when you sit through one of these and

you think you know which way it's going to go, and then, all of a sudden, something comes up and you see it, you know, in a very different light, and that's not -- not atypical.

MS. BERNSTEIN: I think we feel pretty comfortable by the time we get to a hearing because, again, nobody fakes their conditions, it's all a matter of determining the severity. And we're usually pretty comfortable.

But the problem is, until we get to the hearing, in those two years, people suffer so much that if we could get the questions that we would ask at the hearing, whether it's an advocate or whether it's you, as the finder of fact, at that point we wouldn't have the delay because the RFC determination would be made by asking the questions in a way that you'll get qualitative information rather than yes/no.

And I think we also have people -- you know, I'm -- I'm dyslexic, I hate writing on forms. I don't know that I would do such a great job of representing myself if I were disabled, and

I think that that's the information that we're relying on. We need lower level adjudicators to ask the questions that you or I would ask to make sure that we get the real answers on RFC.

COMMISSIONER ASTRUE: We're almost up on time. Let me just do one last check.

Dave, any more?

Frank?

Okay. I think we're all set. And remarkably, we're going to break two minutes early. We've given you a long break, not only because we're aware that people have Blackberries and need work to catch up on, but we are in sort of a restaurant wasteland here, and I -- and I do apologize for that. Not much I can do about that. So we've given a little bit longer than we probably would ideally do.

So we -- we have a break for an hour and a half. We have two more panels in the afternoon.

Oh, yes, there's a list of restaurants in the packet. Best of luck with that.

(Laughter.)

COMMISSIONER ASTRUE: I'm not vouching for any of them.

And we've got two more really interesting panels in the afternoon.

So, again, I want to thank our witnesses for a very helpful presentation, and I look forward to two equally good presentations this afternoon.

Thank you.

(Whereupon, at 12:18 p.m., a luncheon recess was taken.)

A F T E R N O O N S E S S I O N

(2:51 p.m.)

COMMISSIONER ASTRUE: All right. I was having a great conversation, I know panelists and others are doing the same; but as I said, we're trying to embrace timeliness as a virtue throughout the Agency. So I think we should probably get started.

So we're going to move now to our first afternoon panel to talk about rare diseases in adults.

We have Walter Koroshetz, who's Deputy Director of the National Institute of Neurological Disorders and Stroke at the National Institutes of Health.

We have Steve Gibson, who is Vice President of Government Regulations and Public Affairs for the ALS Association.

And Ron Bartek, who is president of the Friedreich -- Friedreich's Ataxia Research Alliance.

Thank you, gentlemen.

I guess we'll start with Walter.

DR. KOROSHETZ: Thank you very much. It's a pleasure to be here, Commissioner Astrue, and members of the committee. So I'm representing the National Institute of Neurologic Disorders and Stroke.

And our mission is to reduce the burden of illness due to stroke and basically hundreds of neurological disorders.

Though some of our illnesses we take care of are very common, such as stroke, epilepsy and headache, we are actually -- also the lead institute for hundreds of rare disorders, many of which affect adults, many of which affect children.

Many, if not all, of these conditions cause major disability in all or a segment of the affected population.

Our efforts are primarily focused on facilitating the discovery of new knowledge and new treatments for these extremely debilitating and, oftentimes, fatal conditions.

Some cause impairments that are obvious, such as paralysis. Others are disabling in much less obvious manners, such as those that affect cognition. Especially difficult are those associated with chronic pain and changes in mental function.

As compared to previous ages in this technological age, one's ability to function in the workplace seems dependent less on physical abilities and much more on cognitive abilities compared to past centuries.

The IOM report entitled, Improving the Social Security Disability Decision Process reported that the percentage of SSI adults age 18 to 64 with mental disorders was 57 percent, and mess -- and mental disability had seen the largest growth in recent years.

Because there are so many rare neurological disorders, we are acutely aware of the difficulty in being expert enough to be able to accurately make disability determinations at all. Because of the complexity of understanding

all of these disorders, clinical neuroscience itself has become more and more specialized.

As a neurologist in -- in a busy practice in Boston before coming to the NIH in January, I had the opportunity to experience the disability system in action, not only in my practice, but overseeing the practice of about 30 to 40 residents in our -- in our program.

And I'm cognizant of the troubles that certain patients have had in determining -- in obtaining legitimate disability determinations. Unfortunately, I can also testify that it was not infrequent to come across a case in which a patient or person perfectly able to make contributions in the workplace would attempt to convert a diagnosis of a neurological illness into a determination -- a -- a disability determination. Sometimes this seemed motivated by fear of their future health, sometimes simply by desire to establish a steady income.

The Social Security Administration definition that a person be unable to engage in

any substantial gainful work which exists in the national economy as opposed to their previous job is also very difficult for many patients to accept.

Physicians, therefore, are often pressured to fill out disability forms for patients who do not fit the definitions of the Social Security Act. In some cases, physicians may object and indicate to the patient that remaining employed is in the best interest of their health. On the other hand, the physician is also acutely sensitive to the importance of the patient/doctor relationship.

They will often, therefore, fill out the form truthfully in these cases, but hoping that the SSA reviewer will see the facts clearly and make the correct decision.

This is not an easy process, I would imagine, for the SSA reviewer. Worse, there are also some patients with neurological symptoms who challenge the best diagnosticians to determine whether they are in fact contrived or due to a

brain disease.

There's some real diagnostic dilemmas that are difficult for the physician to diagnose, would clearly be difficult to make a determination of disability.

So understand that SSA clearly has a tough job being sure that they don't deny benefits to those who are truly disabled and don't grant benefits to those who are not really disabled. In many cases, there is no gold standard.

Troubling as well, as what I mentioned above, is the patient unable to work due to damage to brain, spinal cord, nerve or muscle, who struggles to obtain disability determination. In asking for input in -- into this issue from a number of different disease-related organizations, most of the e-mails that I received outlined the -- the problem as primarily variability and what happens when someone enters the process.

The decisions are difficult to make and when one sees patients with equal disability but different diseases receive different

determinations, it raises concern.

If patients have the same disease and equal disability and also have different experience, it also raises concerns.

The hard thing appears to be to -- to try -- especially with the rare diseases, to try to understand how to make a very consistent decision without -- with the least amount of variability. And it's a very difficult problem, given the complexity of the many neurological rare diseases.

I wanted to just illustrate one example from fronto-temporal dementia. It's a little known cause of dementia. It's been in the paper recently, so people may have heard about it more so lately. It causes degeneration of particular parts of the brain, and it can be variable, depending on what part it attacks. Usually it affects people in their 50s or 60s.

There are multiple different types of frenal -- fronto-temporal dementia, but not as well-known as, say, something like Alzheimer's

disease.

So I received one positive reply, a very positive reply from a -- a -- a spouse of a man with fronto-temporal dementia, and she says, and I quote, my experience -- I'm sorry, it's a -- so it's a -- a -- it's a lady with a mother who has fronto-temporal dementia -- so my experience with helping my mother apply for Social Security benefits was not as hard as I thought it would be. We went to the office together with a summary of the testing information from a neuropsychologist.

At the time, the doctor had not used the phrase "frontal temporal dementia," but instead was described brain atrophy in the frontal lobes, dementia and aphasia. So didn't even give the patient a clear diagnosis.

However, she went on to say that the man at the Social Security office was very understanding and it was obvious to him or anyone that my mother was impaired. There was also the fact that she'd been fired from her job of seven years; then got another job, was fired

two weeks later for not being able to perform the task.

Our meeting was in December, and her benefits started in March. I thought it would be harder.

So this illustrates a good outcome, but it may not be representative of all patients with FTD.

Dr. Kent Jamison, who is vice chair of the Association for Fronto-Temporal Dementia, wrote to me saying that the, quote/unquote, ambiguities inherent to this disorder make it difficult sometimes. Patients can become cognitively impaired at a younger age.

Another person who responded to me with e-mail said, when I called SSA, I was asked if my husband was over 65. When I said no, the reply was, well, if he's demented, why isn't he over 65? This patient, however, also had a very clear pathway through the Social -- through the disability process.

Fronto-temporal dementia affects the

patient's frontal lobe executive function which may be necessary to persist in the pursuit of disability determination process.

Dr. Jamison wrote that many do not even recognize their own deficiencies and, therefore, it's little wonder that the final approval may get tripped up at the local or regional level, causing delays and hardship for the caregivers.

In my first practice, I cared primarily for patients with Huntington's disease in Boston, and I started in the mid-1980s. And it's an example, I think, of how things can change for the better.

When I first started, I recall constant battles in trying to obtain disability for the patients with this degenerative genetic disorder that really destroyed their brain over about 20 years.

Their disabilities were usually due to cognitive changes that made it unable for them to work, but it was not obvious from looking at them

that -- how disabled they were.

But over time, as I filled out more and more, I noticed that by the mid-1990s, there was a complete change. I recall one day thinking as I fill out this form how easier things have gotten in the last 10 years and -- and -- and I think I don't know the answer of why that happened. Was it that Huntington's disease became better known, or the state DDS staff came to know me, or Huntington's was added to the list of degenerative disorders?

I can't tell, but it's an example of how a process which was tough over a period of time became quite smooth and easy for the patients who were really disabled.

The tough issue is how to systematize the knowledge base needed to make accurate disability determine -- decisions in all conditions.

Clearly, the rare conditions present the most problems because an individual's exposure to the wide array of presentations is surely to be

limited.

It's actually what's caused the subspecialization in -- in the neurosciences, because you really can't be an expert in all these diseases. I think a similar thing would apply to the people trying to make these determination processes.

Since I've come to NIH, I've seen that the Social Security Administration has been extremely proactive in trying to problem solve. This summer, representatives from multiple NIH institutes met to discuss how NIH research might help inform SSA with regard to how a new scientific knowledge can impact on the disability determination process.

A number of NIH institute representatives were enthusiastic about participating in the process to uncover whether new technologies, say, neuroimaging, neurogenetics, specialized testing, might be best utilized to determine when a person is really no longer able to work who carries a diagnosis of a

specific disease.

Others were interested in the idea of incorporating commonly used functional or medical severity scales which have been well validated in clinical research arena and potentially could be correlated with inability to work and perhaps inform the Social Security determination process.

All who attended realized the difficulty inherent in making 100 percent accurate determinations on the first attempt in all 2.6 million applicants a year. But they're attracted by the possibility that at least being able to make sure that a significant number of -- of severely disabled patients could be identified in -- with some type of formulaic approach.

The hope is that this might directly help a significant number of affected individuals, improve processing time, but also indirectly help if SSA resources can be better concentrated than on the tougher cases.

NIH is very interested in working with SSA to develop the knowledge base for injecting

the most current and valid mechanisms of determining the severity illness into the disability determination process.

This is a work that is probably always going to be in process as new techniques and information comes out of new research.

So thanks very much for your attention.

COMMISSIONER ASTRUE: We're going to stick to the procedure that we developed this morning and hold off on our questions until all the panelists have spoken, because I think a lot of times panelists -- multiple panelists will want to address our question.

So we'll go to Steve and then to Ron, and then we'll pepper you with questions.

MR. GIBSON: Great. Thanks.

Good afternoon. My name is Steve Gibson. I'm Vice President of Government Relations and Public Affairs for the ALS Association.

I appreciate the opportunity to speak

with you this morning on behalf of the ALS Association and the thousands of people who have Lou Gehrig's disease.

We are pleased to partner with the Social Security Administration as it examines compassionate allowances and other ways to improve and expedite the disability determination process.

The ALS Association is the only nonprofit health association dedicated solely to fight ALS, or Lou Gehrig's disease. Amyotrophic lateral sclerosis, more commonly known as Lou Gehrig's disease, is a progressive, neurodegenerative disease that erodes a person's ability to control muscle movement.

ALS is designated as an orphan disease, with an estimated 13,000 to 30,000 people living with the disease in the United States today.

Approximately 5,000 people are diagnosed each year, and about the same number die from the disease annually. The disease also is difficult to diagnose, and it is often

misdiagnosed, for there is no single test to determine whether someone has ALS. Rather, a diagnosis is made after eliminating other possible diagnoses.

Before the disease -- because the disease is relatively rare, many people are not familiar with the symptoms, its progression or its paralyzing and fatal outcomes. They do not know that once a person develops ALS, their condition will never improve and will only get worse.

As the disease advances, people progressively lose the ability to control their muscles, to walk, move their arms and hands, talk, and even blink an eyelid. Yet, their minds are largely unaffected.

They are isolated and awake, alive with the knowledge that they are trapped inside a body that they can no longer control. Ultimately, the disease robs a person of the most basic human function, the ability to breathe, as people with ALS generally die from respiratory failure because they no longer control the muscles needed to

breathe. This is a horrific disease.

The average lifespan for a person with ALS is two to five years from the time of diagnosis. However, the disease progresses differently in different people, and about 50 percent die within 18 months of diagnosis. This -- there is no known cause, cure or means of prevention for this disease.

While advances in medicine and technology have helped prolong life and improve quality of life by treating the symptoms of ALS, there currently is no effective treatment available that reverses, stops or slows the progression of the disease.

One drug has been approved by the FDA to treat ALS, but that drug, Rilutek, which was approved in 1995, only prolongs life by a few months, and only in some patients.

This disease can strike anyone at any time, regardless of age, race, gender or nationality. However, while we are seeing ALS more frequently diagnosed in younger Americans,

those in their 20s and 30s, the average age of onset is between 40 and 60 years old. In other words, this disease impacts people in the prime of their life, those raising and supporting families, active members of communities, your friends and neighbors, colleagues and coworkers.

The benefits available through Social Security Disability Insurance, SSDI, and Supplemental Security Income, SSI, programs, including access to Medicare, are invaluable resources to people with ALS, and ones on which they rely to enable them to continue to live productive lives and obtain needed healthcare while they are also -- why they also fight their disease.

These benefits are particularly important to people with ALS. The disease generally strikes people in their midlife while they are productive workers of the workforce.

And unlike some other diseases, ALS progresses rapidly and generally is not chronic, providing a family little opportunity to prepare

for sudden loss of income.

Those costs include the cost of medical equipment, like power wheelchairs, b-pap machines and ventilators, speech generating devices, physicians' services, prescription drugs, in-home support services, the handicap accessible transportation, home modifications and many others.

They include the loss of a spouse's income, for they often serve as the primary caregiver, and the additional costs that are required to meet a family's day-to-day needs, needs which were once met by the person now fighting the disease.

It is not unusual for a person with ALS to go from working full-time to being confined full-time to a power wheelchair in less than a year. This rapid change, combined with the significant financial and medical costs of the disease, make it especially important for people with ALS to access Social Security benefits and Medicare in a timely manner.

Until relatively recently, people with ALS have experienced problems accessing Social Security benefits. For example, some people with ALS have been determined not to be disabled simply because they were able to walk into a Social Security office.

The system quite simply did not effectively recognize the progressive nature of this disease. It did not recognize that just a few months after an application was denied or delayed, many of these same people would be confined to wheelchairs, some nearly completely paralyzed, and others no longer are alive.

The system did not know what ALS was and how rapidly it can arrive and -- rob an active, vibrant person of the ability to function; how the disease can disable a person and take from him or her those things which most of us take for granted.

We are pleased to report, though, that in recent years, much has changed, thanks to actions taken by the Social Security

Administration.

In August 2003, the Social Security Administration published new rules that, one, changed the listing for ALS, making it easier to qualify for SSI; and, two, added ALS to the list of conditions that automatically qualify for presumptive disability payments under SSI.

Under the new listing for ALS, only medical evidence demonstrating that a person has ALS is needed to meet the listings and to be found disabled. They no longer had to also demonstrate significant bulbar signs or difficulty using their arms and legs.

In short, the regulations recognized that those things would happen, and the SSA ultimately would find a person with ALS to be disabled.

Importantly, the new rules also enabled people with ALS to qualify for presumptive disability payments simply on the allegation of ALS. This, too, appears to have expedited access to much needed benefits.

As I mentioned, ALS is a rare disease that unquestionably qualifies for disability.

I'm also pleased to report to you that the anecdotal evidence, the experiences we hear from our chapters and the individual patient shows that the system appears to be working for most people with ALS.

In the majority of cases that came to our attention, people with ALS are able to access benefits in a timely fashion. Claims generally are approved quickly, although in some cases, it can take longer than expected and require a patient to take additional steps to demonstrate disability.

While it is difficult to tell for certain, there are a few factors that we believe contribute to problems and delays.

One, the most common problem appears to be lack of familiarity with the disease or understanding of its progressive nature on the part of field offices and state agencies. Unfortunately, that is one of the challenges with

a rare disease like ALS. People may have heard of Lou Gehrig, but they don't know much about ALS.

Two, it appears that in many of these cases, field offices and state agencies are not aware that ALS is included on the list of impairments and that a diagnosis supported by medical evidence is all that is needed to qualify.

Three, other problems include lost or missing paperwork or difficulties in rural areas where there may be less experience with ALS.

Four, additional problems are encountered by people with ALS not related to the disability, but to work requirements like the 20/40 rule and the five-month waiting period.

We recognize that these later issues fall outside the scope of the discussion today and also involve statutory changes.

Ultimately, most of the -- the -- these cases are resolved favorably, and since 2003, we, in our chapters, have encountered fewer instances of difficulties.

It should be noted that there are

several reasons why we believe problems appear to be occurring less frequently and why cases are being resolved favorably.

Since 2003, field offices and state agencies seem to have increased awareness of the rules that apply to ALS. The rules are no longer new.

Second, some of our chapters, as well as the national association, have developed relationships with local offices and SSA staff and conducted our outreach to help educate them about ALS and its inclusion to the listings.

Third, we have provided information and guidance to patients applying for disability. For example, we instruct patients to specifically reference the listings when they apply, contact specific individuals in local office -- offices, and cite the regulatory language as well.

In addition to being included in the TERI listings, the claims of people with ALS also are eligible for expedited process as -- as TERI cases, and individuals -- individuals are eligible

for presumptive disability payments under SSI.

Unfortunately, we do not have conclusive evidence that these processes are either working well for people with ALS or not working well.

We have heard from some chapters that people qualifying for presumptive disability payments do not receive them, not necessarily because the payments are being denied, but because patients or SSA staff simply may not know that people with ALS automatically qualify for such payments.

Again, I want to reiterate that this information is anecdotal in nature. However, we believe SSA should have more definite information that shows whether people with ALS who qualify for SSI also receive presumptive disability payments.

We have a few suggestions.

The anecdotal experience of people with ALS appear to demonstrate that the current process that SSA has established appear to be working for many people.

One improvement we'd like to talk about is educating and training. We believe that it's critical that SSA educate adjudicators about rare diseases like ALS, describing the conditions, their progressive nature and the evidence that is needed to establish disability.

Equally important is reinforcing the rules that apply to specific conditions like ALS and training adjudicators to ensure proper implementation of not only of new SSA policies, but also existing policies.

For example, in addition to being included on the listing of impairments, ALS cases also qualify as TERI cases and also are eligible for presumptive disability payments.

It does not appear that adjudicators are consistently aware of these facts. In some cases today, applicants are the ones educating adjudicators about new and existing policies.

Community outreach. We encourage you to work with us to reach out to rare diseases and the disabled community and to enlist our

assistance in serving these constituencies. Indeed, we welcome opportunities to educate local offices about ALS and to provide information to them that would make their jobs easier and reduce problems to expedite claims.

Three, educating the rare disease community. While SSA has available a variety of publications to inform the individuals of disability benefits, their rights and applying for disability, we are not aware of publications or information that is more targeted to those with rare diseases, like ALS, or those who unquestionably qualify under SSA's listings.

We believe that making such material available to individuals and organizations could help to further improve the current system and enable them to more easily navigate what can be complicated and confusing processes.

Technology. We applaud SSA's initiative to utilize technology to improve the speed of the disability determination process and reduce the prevent -- reduce and prevent backlogs.

The quick disability determination, QDD, process is an excellent example of this in which SSA uses computer screening tools to rapidly identify cases that are likely to qualify.

We encourage SSA to continue to examine in and of -- innovative ways like this to apply technology in all facets of the application and review process.

Streamlining applications. Finally, we believe that for those individuals with conditions that unquestionably qualify under SSA's listings, you consider streamlining the application process that is required to apply for disability or to support a claim.

Some of these current requirements, such as indicating the number of steps one can take, are not relevant to people diagnosed with ALS. In fact, they may deter people from applying for benefits or pursuing a claim if they erroneously believe they will not qualify for benefits.

We believe SSA should consider

processes that permit people with ALS and others who unquestionably qualify under SSA's listings to avoid submitting information that may be required in most cases, but is not relevant to their application. For example, applicants should simply be directed to skip certain sections if they have a specific condition like ALS.

Thank you for providing me with the opportunity to speak with you this morning. The ALS Association applauds your efforts to improve the disability determination process for people with ALS and other rare diseases and disabling conditions.

COMMISSIONER ASTRUE: Thank you.

We'll move to Ron.

MR. BARTEK: Good afternoon, and thank you, Mr. Astrue, for -- and the members of your panel for undertaking this tremendously important and tremendously complicated undertaking of improving, wherever possible, the adjudicatory process and the SSA benefits as they pertain to patients with rare diseases.

Thank you, too, for holding this series of hearings and -- and for inviting all of us to speak.

I'm Ron Bartek. I'm president and co-founder of the Friedreich's Ataxia Research Alliance, or FARA. We are a 501(c)(3) nonprofit organization devoted entirely to supporting the research into Friedreich's Ataxia.

And I'd like to say that that research, that body of research is so promising right now, not only in terms of its promise for providing and developing treatments for Friedreich's Ataxia, but also, as we realized only over the course of the last year or two, providing powerful insights into and potential benefits for patients with other diseases, some rare, not so -- and others not so rare, like Parkinson's disease, Huntington's disease, Alzheimer's, ALS, stroke, diabetes and Alzheimer's disease, a series of rare mitochondrial dysfunction disorders, like MELAS and MERRF and Leber's Hereditary Optic Neuropathy.

So we are wonderfully excited in our

community about the prospects for treatment.

We're getting five clinical trials, maybe even six over the course of the next 12 months. And we are absolutely convinced that insights gained and benefits gained from those clinical trials will be beneficial for other disease groups that you're dealing with today and in your work.

But on -- on the core topic on which I'm asked to address the panel, I've been asked to tell you just a bit about our family's experience with the Social Security Administration's benefits for rare disease.

And so let me -- let me say in that regard that our son, Keith, has a rare, life-shortening genetic disorder called Friedreich's Ataxia.

Though Keith was born a healthy infant, as was the case with many of the panelists today, in December of 1985, by the time he was 8 years old and in the 3rd grade, he began to stumble and fall for no apparent reason.

He also began to have increasing difficulty writing legibly. His hands were cramping and so were his legs. These symptoms worsened to the point that when Keith was 11 years old, we had him tested, and he was genetically confirmed to have Friedreich's Ataxia.

Like most parents who receive this diagnosis, we had never heard of Friedreich's Ataxia. I wish I could still say that. We learned a lot about the disease, though, on the day of diagnosis.

We learned that Friedreich's Ataxia is a neuromuscular disorder inherited by way of a recessive genetic mutation carried by about 1 in 90 people. We learned that when two such carriers of -- have children, each offspring has a one-in-four chance of inheriting the mutation from both parents and being afflicted with the disorder.

We learned that Friedreich's Ataxia is relentlessly progressive and would soon deprive Keith of strength and coordination in all four

limbs, placing him in a wheelchair by the time he was a teenager.

We learned that Keith's vision, hearing and speech would all be diminished; that he would develop severe scoliosis, or curvature of the spine, requiring surgical implantation of metal rods to straighten it, a horrific and brutal surgery; that he would be at greater risk of developing diabetes; and that he would most likely have a severe heart condition.

Finally, we learned that there's no treatment or cure for Friedreich's Ataxia, that average life expectancy for Friedreich's Ataxia patients is early adulthood.

Within -- within just a few weeks of Keith's diagnosis, after a number of medical examinations, we knew he already had a severe heart condition and the early stages of scoliosis. His weakness and incoordination progressed rapidly, and he began using the wheelchair full-time when he -- when he was 16.

His scoliosis also worsened, and we --

and he required the spinal fusion surgery to correct his spine that same year.

His vision, hearing and speech steadily declined and continue to do so. He developed Type 1.5 diabetes at age 20.

So by the time Keith turned 18 and was eligible, or we hoped eligible to apply for SSI as an adult, he had been in a wheelchair for more than two years, had endured spinal fusion surgery, could not speak clearly, and his vision and hearing were both impaired.

He had an individual education plan for the final seven years of his education. Keith had worked hard and done well in school, despite his disabilities, and graduated from high school with his class after posting a 4.0 grade point average during his senior year.

Although he went to our local community college for several semesters and did well in his classes, the relentless progression of this disorder left him extremely fatigued and made it increasingly difficult to read and keep up with

his assignments.

Transportation was another complicating factor in that Keith had never been physically able to drive, so had to rely on others to get him around.

With his physical abilities and the steep decline and the demands of his education on the rise, Keith reluctantly decided he could not continue his schooling.

He was also disappointed to find that he was unable to find a job.

Again, because Friedreich's Ataxia is relentlessly progressive, Keith's physical capabilities have continued to decline steadily to the point of needing around-the-clock assistance for even the most basic needs.

The diabetes associated with Friedreich's Ataxia carries its own additional requirements in this regard. Keith is unable to perform the blood glucose testing or administer his own injections, for example, both of which are required several times each day.

With all of these increasing challenges, you can understand how welcome to our family was the very quick SSA decision on Keith's disability claim. You can imagine how encouraged Keith and we were when he received, for example, his first SSI check even before he re- -- received his approval letter.

With no other income or resources, Keith had no other way to support himself. The SSI monthly payment helps Keith pay for rent, food, utilities and personal care items. It is critical for Keith to have the financial ability to take care of his basic needs.

Fortunately for Keith and us, Friedreich's Ataxia is one of the rare disorders listed in Section 11.17 of the disability evaluation under the Social Security Blue Book of June 2006. Because of Keith's positive experience, he and we now realize that not only is the SSI program extremely important in and of itself, SSI approval also provides a gateway to applying for additional social services, for

example, Medicaid.

Because Keith was approved for SSI, he was able to apply for Medicaid through the Commonwealth of Virginia. He met the Federal income and asset eligibility standards, and Medicaid helps pay for his doctor and hospital care, medication and durable medical equipment, such as his wheelchair.

Home base care. Keith has continued to decline dramatic -- dramatically in his ability to take care of himself. Personal grooming, dressing, meal preparation and eating, just to name a few tasks, he now finds very difficult or impossible.

With SSI and Medicaid approval in place, Keith was evaluated and found eligible for personal care services. He receives caregiver services five hours per day, Monday through Friday. The caregiver helps him bathe, dress, prepare his meals, washes his clothes and vacuums his floor -- vacuums his room. Not only is this of benefit to Keith, it allows us, my wife and I,

daily -- a daily block of time for our own employment and time to care for our other family's routine needs.

Respite care. When Keith's caregiver is not with him, either his mom or I have to be available to take care of his needs. This reality has become part of our daily lives. We love our son and want to help him in any way we can, but we realize at the same time, like so many others, we will not be able to help take care of Keith unless we take care of ourselves and our other obligations.

Keith and we were grateful, therefore, that Keith was granted approval of preauthorized service for respite care in the amount of 720 hours per year. Respite care helps to refresh us, to sustain ourselves and our family. It helps us take care of ourselves and our family so we can take better care of Keith.

I doubt, Mr. Commissioner, that anyone will come before you and say that it is easy living with a rare disease or helping care for

someone with a rare disease.

I would like to assure you, though, that the SSI program helps many people, including our son, both directly and indirectly, in tangible and intangible ways.

It helps directly by providing him the wherewithal to provide for himself as much as possible. It helps indirectly by -- by providing a valid foundation that opens the door to important additional social services.

It helps in the tangible physical ways I've outlined. It helps in important intangible ways, too. It reinforces the recipients self -- sense -- sense of self-sufficiency, self-esteem and dignity. It works in all these ways to help provide the recipient with a higher quality of life than otherwise could be achieved.

I applaud the Social Security Administration in reviewing the need for compassionate allowances in an attempt to streamline this system for other patients with rare diseases, and I look forward to assisting in

any way I can.

In terms of your efforts to develop the most effective methods for identifying rare medical conditions appropriate for compassionate use allowances, I know that you're already engaged in most, if not all, of the following undertakings, so I will simply emphasize them for the vantage point of our own very positive experiences.

One, I would suggest, for example, continuing to work closely with your colleagues at the National Institutes of Health, obviously, as the membership of this panel indicates you are doing.

More specifically, I would suggest that you continue to work closely with the NIH Office of Rare Diseases, represented so ably by Steve Groft, as well as with each of the NIH institutes responsible for each of the rare diseases under review.

Again, the list of witnesses for this hearing makes clear that you are on that case, so

to speak.

I had the good fortune to serve for four years on the National Advisory Committee of one of those NIH institutes, the National Institute for Neurological Disorders and Stroke, represented here on this panel by its deputy director, Dr. Koroshetz.

And I would like to join the millions of others in applauding the NIH as the world's leading funding agency for medical research. You are no doubt finding at the NIH highly qualified scientists who can help identify the trigger point, so to speak, within rare diseases that can serve to validate speedy SSI approval.

If there is anything you believe a patient advocate like myself might be able to do to be of assistance in this regard, in a liaison capacity, for example, I and many others would be happy to work with SSA in any way that might prove helpful.

As president of the Friedreich's Ataxia Research Alliance, I would also like to

suggest that you continue to work closely as you are with nonprofit advocacy organizations that support research and services for patients with rare diseases.

Such organizations have formed around many rare diseases and would be happy -- more than happy to help wherever they can.

The National Organization of Rare -- Rare Disorders, NORD, which was represented ably this morning on a panel, is represented on -- is also clear -- clearly ready to help.

To illustrate the same point using our own organization, FARA, we frequently answer questions and provide advice to patients who are preparing to file an application for SSI. We help them, for example, in -- in identifying the employment, education and medical documentation required for filing with the other Social -- specific Social Security regulations pertaining to our disease and in using the online screening tool, called BEST.

Rare disease patient advocacy groups,

as you pointed out in front of an earlier panel, tend to know a lot about their disorders in part, as you pointed out, because they have to educate themselves as well as many of their family physicians who didn't learn much about these rare diseases in medical school and have had precious little contact with them since.

Such advocates and -- and the scientists with whom they are closely aligned could offer significant help in identifying the trigger points for compassionate allowances and these rare diseases, and they can be extremely helpful in educating the rare disease patient community in terms of the process involved in applying for SSA benefits.

For example, going back to the excellent exchanges you've had this morning with previous panels, the medical advisors working with such advocacy groups could provide invaluable insights and potential function -- on potential functional criteria. In the case of a number of rare diseases, for example, clinical networks that

they've helped develop -- that the advocates have helped develop, are working hard to develop clinical measures and scales essential to determining progression of disease, as well as therapeutic effect of any therapy to be tested in a clinical trial.

These measures and the scales behind them are fine tuned to the specific disorders in question and would include validated questionnaires for quality of living and activities of daily living.

Like the -- the MELD test that -- or scores that you mentioned this morning, Commissioner Astrue, for -- for liver, there are such scores being developed for not just individual rare diseases, but in many cases, for collections of rare diseases that answer to the same underlying mechanisms of damage and, therefore, might respond to the same underlying mechanisms of action for therapeutics.

So I think that's a really promising avenue of approaching the advocate, the NIH,

the -- the rare disease groups and their scientists and advisors, because some of those measures and scales and -- and scores are being developed.

In many cases, the request -- first, currently, the SSI application process requests very generally medical reports, and in many cases, that request for general medical reports is adequate. It wasn't ours, because we had -- we know our gene, we know -- we have a genetic confirmation test, we know a lot about our -- our symptoms, and it's relentlessly progressive. And, in most cases, the applicants for SSI are already in their wheelchairs. You know, they're already clearly able to qualify under the single functional capacity which is, in our case, quote, significant and persistent disorganization of motor function in two extremities, resulting in sustained disturbance of gross and dexterous movement or gait and station, end quote.

Our son was already in his wheelchair, as most of your other applicants are; he was

already unable to communicate clearly, clearly met those -- that functional capacity criterion.

However, in cases of later onset of our disease -- there are cases of later onset, so that the adult doesn't develop the symptoms until mid 20s, sometimes even later, that would not be so easy a call.

And so in that case, the process that you've already outlined of consulting so broadly, as you already are doing, can further instruct the -- the process and -- specifically by asking more -- for more and better information at the outset, more and better information in the early stages, rather than waiting for the hearing stage two years later.

At the application process, if -- if the list and the functional criteria -- criteria are instructed by the kinds of things you will learn in these consultations, we believe that -- and -- and maybe even the measures and scales and -- and test scores that -- that -- that you're looking for are there, available to the very

helpful local representative of SSA -- in our case, extremely helpful, sat down with us, helped us fill out the application, gave us some guidance on where to go to get these medical reports.

But if -- if that local representative was -- had the additional information in front of her or him to be able to tell the parent and the patient, you know, this is the kind of medical report you need and -- and if you've -- if you've gone to a clinic that has a specific examination for your rare disease, can you get a test score?

And -- and, you know -- those -- those test scores are increasingly available in a -- in a -- in a number of rare diseases like ours, and I think that will continue to be the case as you do your work.

So in -- in -- in sum, I -- I believe the kinds of collaborations you are already discussing with -- with your panels and -- and represented by your colleagues on your own panel and -- that productive collaboration is -- is already afoot and -- and that you will benefit

tremendously and that -- that collaboration will be extremely productive and result in more and better information early in the process.

In final conclusion, thank you very much for the benefits of the SSI program. They provide to our -- our son and many others in our families a -- a great deal of benefit, both tangible and intangible.

Thank you also for your efforts to make SSI available quickly to the other families living with such devastating rare diseases through the proposed compassionate allowances initiative. And it's my pleasure and honor to be here.

COMMISSIONER ASTRUE: Thank you. Thank you very much. Thank you to the entire panel.

I've got a -- a few questions here.

Both these diseases are ones that I worked on professionally in the past, and so, you know, I don't have to be persuaded on, you know, what these disease are like.

So I've just got a couple technical

questions.

I wanted to ask you about ALS in terms of the difficulty of diagnosis. And if you could maybe walk me through a little bit what your experience has been in terms of what it tends to get confused with and then how that gets sorted out, because if we know that, that might make it a little bit easier for us to be clearer in our standards when we're trying to sort that out to make sure -- because the question for ALS is really just do we have a legitimate diagnosis?

As you know, you're in -- ALS is in the QDD program now, and last week I think we were up to 28 states, we should be 50 states before very long, and they should be routinely allowed within sort of 10 to 14 days very shortly. So, I mean, there is some real progress going there.

But it seems to me in the case of ALS, the big progress is just making sure that we're asking the right questions to make sure that we've got the right diagnosis.

So if you could walk me through that a

little bit --

MR. GIBSON: Sure.

COMMISSIONER ASTRUE: -- that would be helpful.

MR. GIBSON: And we'd be happy to provide the Agency with a more medical/scientific background. I'm a government relations professional, I can tell you what I've experienced in my 10 years of the office here in Washington.

As I said in my remarks, there is not one test, so basically a person with ALS goes through a series of -- of tests and various doctors' appointments.

We've had stories of patients coming in, first being diagnosed as carpal tunnel syndrome, where they actually have not had movements in their hands. We've had situations where it's confused with Lyme's disease, where there's some connections.

COMMISSIONER ASTRUE: Probably Guillain-Barré and fibromyalgia and things in that class, too?

MR. GIBSON: Right, right. Correct.

And sometimes there is some confusion with MS. Generally what happens is ALS is -- is a death sentence. So any sort of medical professional does not like to give that diagnosis until everything else is ruled out. But, you know, a lot of times -- we have stories of an actor who was on a soap opera who was fired from his soap opera because they thought he was actually using drugs because he couldn't negotiate certain words, and that went through a whole legal ramification with Proctor and Gamble where they finally diagnosed him with ALS, and that took two and a half years.

But we'd be happy to give a list for all the agencies to look at for various examples.

COMMISSIONER ASTRUE: Yeah. Well, maybe you -- you can update us.

DR. KOROSHETZ: Yeah, it's a -- you know, there's -- like in many different neurological diseases, there are a variety of presentations, and the classic ALS presentation of

weakness, atrophy of muscles, vesiculations in the muscles with confirmation with electrophysiological test, the EMG, you can make a fairly significant, you know, diagnosis.

You'd like to exclude, of course, anything that's treatable because of ALS not having a treatment. There are some disease -- disorders that can mimic ALS, but those -- I think those kind of sort themselves out.

The ones that are tricky are the ones -- as -- as Steve mentioned, for instance, do they present bulbar-wise. So it's primarily speech, and there is no weakness, so somebody is saying, well, how could you have ALS if you're not weak, and there's a bulbar form that you have to know about.

Or there's another form, which is progressive lateral sclerosis, where the primary problem is actually in the motor cortex, and the patient develops incoordination and stiffness but not that much weakness. So those are the kind of variations that sometimes will cause someone not

to be diagnosed.

There's another form where it affects the respiratory muscles, and they present with shortness of breath. Sometimes they end up in a medical unit because they're having trouble breathing, and that -- that can be a real tricky one to sort out.

COMMISSIONER ASTRUE: Is -- is there a time -- now the -- the time -- usual time from diagnosis to death is just a few years, usually, right? Is there -- from the time where there -- there -- there's an attempt to diagnose to when you really have a -- a confirmed diagnosis, does that tend to be three months? six months? twelve months?

DR. KOROSHETZ: You know, it's on an average -- you know, my rule is try never to make that kind of diagnosis the first time, always give a chance to see what happens over time. I'd say an average is somewhere around 3 months if you're in a -- you're in a system with good neurology.

MR. GIBSON: But I think that varies

on where you live in the U.S. If you're in a rural area where you might not even have a neurologist in the vicinity, you have to then travel on.

We have, you know, many folks in the Dakotas and Montana have to go to Mayo, so I think that can take months to get another appointment, which then leads to a year and a half or so.

COMMISSIONER ASTRUE: Okay. I had another -- another question for Ron, and again, I -- I realize, consistent with Steve's response, I may be asking too much medical, and so if -- if there's anything you want to submit for the record afterwards from the -- from the Alliance, I'd be -- be happy to take it.

I have some general recollection that Friedreich's Ataxia is sort of the most common in sort of a loose family of somewhat similar diseases, most which of are even rarer than Friedreich's Ataxia.

Could you may -- tell me maybe a little bit more about the -- the other ataxias and

whether you have any observations about the problems that they have in terms of diagnosis and system -- and -- and -- and going through our system and other systems as well?

MR. BARTEK: I will give it my best shot, Commissioner Astrue, and -- and -- and provide some additional materials later.

But, yes, you're referring to a general family of ataxias, and there are on the order of two dozen or a few more now, maybe up to 30 different identified ataxias. The vast majority of them are dominant trait ataxias.

There are only probably two or three recessive trade ataxias like Friedreich's Ataxia and Ataxia telangiectasia. And the -- and, yes, you're right, Friedreich's Ataxia's is by far the most common of those ataxias.

Roughly speaking, Friedreich's Ataxia patients constitute about half of the ataxia in the population, all the others combined being about an equal number.

They -- about -- I'll venture a guess

that there are roughly a third to a half of those ataxias now that have genetic confirmation tests available, and leaving the other half without any specific genetic marker. And the majority of those with specific genetic confirmation are, like Friedreich's Ataxia, triplet repeat expansion diseases, polyglutamine ataxias, the difference being that those triplet repeat expansions are on coding regions, exons of their genes, so they -- they -- they do a gain of function problem so that the protein produced by those genes are problematic.

In our case, our expansion, believe it or not, is an intron, and so every time we get a pass, an effective pass, we get effective protein, we get a perfectly formed frataxin protein. So that introduces differences in therapeutics, obviously.

But -- so that -- that's a -- an attempt at summarizing an answer to your question.

COMMISSIONER ASTRUE: What --

what would probably be helpful to us, I -- I -- you know, I know that our folks are -- are -- are on top of Friedreich's Ataxia. What I don't know is, for the whole rest of the family, exactly where they are and if they're up -- you know, up on definitive diagnosis and that kind of -- if -- if you could submit --

MR. BARTEK: Yes.

COMMISSIONER ASTRUE: -- that for the record. Again, it may be that I'm not giving them enough credit, but it certainly wouldn't hurt to have the most recent analysis. If you have that, that would be great.

MR. BARTEK: I -- I -- I will do that, and I'll say at this point that there's a great deal in common in terms of symptoms. Ataxia is a Greek word, as you well know, meaning incoordination, and there is plenty of incoordination amongst all two dozen disorders.

A key difference, though, is -- is age of onset. For the most part, the dominant ataxias, the spinal cerebellar ataxias, are much

later onset. Our onset is usually between the ages of 5 and 15. There are -- there are these cases of later onset Friedreich's Ataxia where the onset can be in the 20s, 30s, maybe even later. So --

COMMISSIONER ASTRUE: Is -- Ron, is there any -- can you tell from genetic testing between the -- the early onset and later onset or do they appear the same in the gene?

MR. BARTEK: There are some markers that are generally true. For the most part, you know, again, it's -- these are triplet repeat expansion disorders, and the size of the expansion in the shorter allele is going to be usually indicative of age of onset, speed of progression, severity of final outcome and nonmandatory symptoms. Nonmandatory symptoms include the heart condition, diabetes and -- and so forth.

So the lower the expansion, you know, the shorter the expansion in Friedreich's Ataxia, because it is in the intron, the -- the assumption is the more frataxin protein you get, and so

the -- the later the onset.

So that could be a helpful, you know, sort of designator or marker, if you will, for the later onset patients that will eventually -- it is relentlessly progressive, so at some point, they will be eligible in almost every case. But I know you'll have to determine, you know, when -- when the onset is sufficient to -- to trigger the benefits.

COMMISSIONER ASTRUE: And that's a good point. And that actually triggered me to -- to just sort of say something, so I'm not going to pretend that everything's a question.

One thing that is important for anyone in the audience that is trying to help us help you to remember is what we're -- we've got this push to look at imaging data and biomarkers and -- and try to draw distinctions in that way.

The statute also is very specific that we're actually not only allowed, but required when -- when it's appropriate, to use age in making these -- these types of judgments, too. So

in many cases, many diseases, that's not very helpful, but there are certain degenerative diseases where the natural history is arguably so predictable that we might be able to use age as a factor, too.

So if you're -- you're thinking a -- about ways to help us draw the line in an appropriate place, don't over -- don't overlook age, because there might be -- probably relatively few diseases and conditions, but there might be a few where the natural history is sufficiently consistent that we might be able to use that as a fuller or partial factor in a decision.

All right. I've been hogging air time.

Anybody to my left have a question?

DR. GROFT: Yeah. I -- I've got a question for Ron or Steve. You both have a considerable amount of information available about the individual diseases that -- that you represent. And I think we heard this morning in earlier panels about needing more information,

maybe different information.

I guess the question is: How difficult, from what you've heard today, would it be -- and, Ron, you've done through the process personally -- how difficult would it be to recast the information that you currently have available and to make it information that would be useful to the Social Security Administration?

MR. BARTEK: I don't think it would be difficult at all, Steve, and we'd be eager to do that. We -- we are fortunate in having a clinical research network already established, a network that's already seen several hundred of our patients, have developed over a three-year period natural history progression against these ataxia scales, if you will. That includes such things as a 25-foot walk, with or without assistance, you know, a time to walk, nine-hole pegboard test, vision testing and speech testing and -- and so forth.

So these scales are readily available. We could provide information that -- that's a

national history for these patients -- that -- that set of patients that I think would be extremely helpful.

MR. GIBSON: Yeah, it would not be difficult at all. Back in 2001 -- actually, 2000, when Congress passed the Medicare waiver for people with ALS, we started to develop a partnership with -- not only with the SSA here nationally, but throughout the country, and we'd be happy to continue that partnership on giving the information.

COMMISSIONER ASTRUE: I should -- I should note, too, one -- you know, one of the reasons why we're drawing two categories, the compassionate allowance and the QDD, for a lot of the people that are very compelling, we can't make a complete judgment, so we have to go through our full, you know, process to evaluate function.

One of the advantages of having the compassionate allowance category -- and, again, we -- we haven't made final decisions on our business processes and forms, and -- and that may

be a work in progress for some time, but it at least opens up the possibility that we can do a short form since -- since really, for compassionate allowances, if we're operating on a -- on a conclusive presumption, all we really need to do is confirm that someone is eligible for Social Security and whether they're Title II or Title XVI and -- and confirm the diagnosis.

We don't necessarily need all the other information that's going to be relevant for the decision.

Likewise, in most, but not all, states, we have rules that require both an examiner and a medical professional to review a case. We have, in some states, what we call single decisionmaker. If we're operating on basically a conclusive presumption, we also ought to be able to cut out one level of review, and -- and I -- I would think go to single decisionmakers, too.

And that's -- that is something that we've heard from a lot of the states which are

really genuinely enthusiastic about this whole endeavor.

So both of you, I think, have touched on it briefly, and I just want to say, we are trying to be responsive to that. I don't know that we -- with all the systems changes that we have to make and all the pressure we're under, I'm not sure that we'll have that the way we'd like to have it eventually when we try to do the first rollout next year. But it's certainly our aspiration to make this as -- make application, particularly for people on compassionate allowances, as easy as possible.

So -- David, do you have anything, or --

ACTING DEPUTY COMMISSIONER RUST: Just two quick ones.

Mr. Bartek, you had mentioned -- and this is something you'll probably want to submit to us as -- as opposed to trying to answer today -- but you mentioned that your -- your organization or organizations are beginning to

develop numerical metrics.

We'd be very interested in seeing where that is and what you've developed, so if you could submit some information on that, I think that might be very helpful to us in crafting this initiative.

MR. BARTEK: Thank you. I'll -- I'll look forward to doing that, and it won't take long because we -- we've been developing these metrics for about nine years and -- and we've worked very closely with the NIH in doing so, and we even have them in electronic form.

So it's -- and we -- we have what we call a FARS score, which is comparable, in some ways, to the MELD score that the Commissioner was talking about. And -- and we -- there will probably be a numeric metric there that says if you -- if your FARS score is greater than 22, you know, you're eligible.

You know, I -- I think --

COMMISSIONER ASTRUE: That -- that potentially could be important. I mean, obviously

not just for your disease, but --

MR. BARTEK: Exactly.

COMMISSIONER ASTRUE: -- for a whole wide range of people. So we'd be very interested in seeing that.

MR. BARTEK: Yeah.

And, in fact, let me say that in that regard, that large portions of that scale that produces that score were -- were borrowed from the MS scales, and portions are shared in common with the Huntington's disease community and -- so I think there -- this will be a pocket of disorders. And including all the other ataxias, you know, that at some point, you know, these kinds of scales will be productive of -- of a score that will be very helpful.

COMMISSIONER ASTRUE: Good.

Yeah.

DR. KOROSHETZ: I'd just say -- that, you know, in trying to develop treatments for diseases, the first step is to develop a scale whereby you can follow patients. So these scales

are all kind of built out of consortia that get together to try and develop a method of knowing if a disease is affected by a treatment. And some of them, like in Parkinson's, there's probably 10,000 people who've been scaled up, or Huntington's.

The rare diseases are -- may not have the same numbers, but any disease where you're trying to get a treatment, they're going to first have to get a scale, and the issue is can you then correlate a score on the scale with inability to work.

COMMISSIONER ASTRUE: Right.

DR. KOROSHETZ: And, you know, if you've done 10,000 people, the -- the data is out there somewhere. If -- you know, if we can capture it, it might be a way of going quickly to a disability determination.

COMMISSIONER ASTRUE: And -- and we have -- we've stopped a couple of things in process. We've got a little room in our R&D budget. We've never actually run studies like that before to try to correlate those types of

data with functionality, but we're, as you know -- you know this because we've been hounding you, but most people in the room don't know that we're very keen to try to start doing this, and we realize to do it right, we may have to do some investment in our -- from our R&D portfolio, which is a new practice for us, but we're -- we're committed to doing that.

So --

ACTING DEPUTY COMMISSIONER RUST: And -- and we also have the ability to pull cases and look at what the outcome of the -- of the -- the -- the determination process was vis-a-vis those scores and give us some idea of how those scores correlate to disability, to a determination of disability, so --

I was also going to thank Dr. Koroshetz --

COMMISSIONER ASTRUE: You may -- you may need to get closer to the mike.

ACTING DEPUTY COMMISSIONER RUST: Excuse me. I also want to thank Dr. Koroshetz for

his -- his help in -- in getting this initiative off the ground and just ask you, you know, in our discussions in August, when you had a number of your colleagues, there was some interesting ideas that were surfaced again about the idea of -- like with schizophrenia plus a cognitive test, the -- the diagnosis plus a cognitive test might -- might lead to a determination.

I was just wondering, are those discussions still going on? Have you been engaged in any of those around the office?

DR. KOROSHETZ: No, I think, as you said, the -- the issue of -- I think is what the common theme that's -- a lot of people come up with is, you know, the diagnosis is not enough, and in some cases, it is, as you mentioned, Commissioner Astrue, the diagnosis determines disability.

In a lot of cases, it's a -- it's a match between the diagnosis and the severity of illness, and the question is then how do you measure severity of illness?

The Social Security forms that the doctors fill out is kind of a generic way of doing it, and to -- to the issue that I brought up is that if you -- you know, if you want to go into the rare diseases, you get so far -- so far with the generic determination, but you may need to -- you know, to have a -- a -- a -- a specialized view towards some of these rare diseases, and that's when these disease-specific scales can -- can come in.

Now, in the -- in the -- in the more common diseases, you know, when I -- when I've seen patients who have gotten disability who I did not think were disabled, it was usually because they had the diagnosis nailed, but the severity of illness was -- was not recognized by the -- by the adjudicator, I guess was assumed to be more severe than it was.

So I think that -- that's the tricky part, I think is -- is being transparent about what is the severity of illness that's associated with disability to eliminate your false positives,

but to put that into a process so it's not so tricky to negotiate that you end -- end up, you know, failing a lot of patients who really are disabled, but they just didn't get the scale measured right or something peculiar about that scale in their position.

I think those are the kind of things we have to try and work out. And you need data to be able to know, you know, your sensitivity and specificity of any kind of a -- any kind of a test like that.

ACTING DEPUTY COMMISSIONER RUST: Thank you.

MR. BARTEK: If -- if I could take the liberty and just expand on that very important point. Dr. Koroshetz is talking about the difference between a generic assessment, which in our case would be a genetic assessment, and -- and severity of illness at that particular point in time when eligibility might be an issue.

And -- and going back to the kinds of scales, Dr. Koroshetz is, boy, completely accurate

in our experience that you can't even pretend to be preparing for clinical trial if you don't have scales. FDA would laugh you out of the room. And those scales --

COMMISSIONER ASTRUE: Sadly, I've had that occur.

MR. BARTEK: Yeah, right.

And those scales include not just the -- the measurements that I've described, like 25-foot walks and pegboard tests and so forth, but really, a -- a quality of life -- living, you know -- a score and activities of daily living score.

You begin with the standard forms, like SF-36 for quality of living scales, but then they're made specific to each of these diseases in -- in the clinical experience that -- that after you've seen several hundred, or in some cases 10,000 patients, you know what questions to ask.

Going back to another discussion you had this morning, you're not always -- the SF-36

doesn't always ask the right question for this particular condition. So now these wonderful clinicians are refining those and adding questions to the SF-36 and so forth. So these scales now include that.

So I think they can be wonderfully instructive in your task.

COMMISSIONER ASTRUE: Okay. Frank, do you have anything you want to ask?

JUDGE CRISTAUDO: I -- I would like to -- to ask Dr. Koroshetz. I'd like to follow up on a point that he made in his testimony in following up, actually, on a question that -- that David asked about.

You mentioned, Doctor, that the doctors who have to complete these forms in -- in practice -- and you were in practice, it sounds like, for many years -- feel pressured, there's kind of a dilemma that they're faced with in -- in completing those forms, and it's obviously a critical issue in -- in terms of what the severity is and -- and the way we make our decision on --

on disability.

And -- and I've asked this question of the earlier panels, and I'm -- I'm trying to determine, is there a better way for us to ask the -- the medical sources for their opinion on what the functional limitations are that a -- that an individual has as a result of the impairments?

I know in many cases we send out a form that lists, you know, how -- how long can they sit, stand, walk and that sort of thing, and there's some thinking that perhaps an alternative approach would be better to ask in terms of limitations imposed by the -- by the impairments.

And I just wonder if you had any thoughts on what would be best for the medical sources.

DR. KOROSHETZ: So I thought about that and, you know, the -- the best way to solve the problem would be to have -- and this is just my guess now, and I have to -- maybe I'm wrong, I have to think it through, but my -- my best guess is that to have an objective measure of severity

that is really transparent is probably the easiest thing to do, because -- because -- because nobody -- you know, the -- the -- the -- the physician wants to do the right thing, and I don't think that they ever fill out the form untruthfully, they put down the information that's asked and -- and then the question -- and then -- and their issue is, okay, I put it down and let's see what SSA does with it.

And so the -- the issues that -- if -- if there was a transparent scale where, you know, you had followed this patient for four or five years, you're going to follow them another five years and you -- you scale the disability, their -- their severity score, that that is a red flag when the patient is getting disability and their severity score is minimal to nothing.

I think that would probably be the easiest way to go. Because it's an objective thing, it's not something somebody can argue. Someone has aphasia, so you put down aphasia, you know, and aphasia, define inability to

communicate, to discuss things with coworkers, and -- but the -- but you never -- but there's no way in the form to actually indicate what the degree of severity of the aphasia is.

So -- so I've seen patients who have an aphasia, no question, you put in the form, they have an aphasia, they had a stroke, they've recovered significantly and -- but you don't have -- there's no way of saying how -- how bad that aphasia is. The aphasia scales which are out there are not -- are not -- not asked for.

So no one knows -- so the diagnosis is there. The patient gets disability because they had aphasia after a stroke, but what no one knows is the aphasia is really pretty minimal, and they actually do pretty well. They can't -- you know, usually in these instances, they can't perform the job that they had before.

So a guy who was a -- you know, a CEO, has a stroke, he has an aphasia, you know, he could -- he could work at another level, he can't -- probably can't work as a CEO anymore, but

that's -- you know, those are the kind of problems that -- that the Social Security Administration doesn't actually get a good sense of the severity of each of the issues.

I think a scale of -- severity scale in my mind would be, you know, a red flag when things are not -- not -- don't seem to match, severity is low. And also, you know, potentially and automatic when you have a disease, as you well know, and you hit this level of severity, everybody knows we have 10,000 patients, you hit that level of severity, everybody's -- nobody can work, that would be another advantage.

Does that make sense?

JUDGE CRISTAUDO: It does.

Like having a GAF scale, for example, but for physical impairments --

DR. KOROSHETZ: Yeah.

JUDGE CRISTAUDO: -- I think is what you're saying.

DR. KOROSHETZ: Right. The -- the issue is -- to make -- for patients -- and I guess

what we're getting at, and this is a tough thing to swallow, is that these -- that the -- that the disease is so different that to have a generic scale gets -- leads to loopholes, I think. To have a more specific scale for the disease is probably the better way to go, but then that's an incredibly complicated thing to put into place. That -- that's where I see the attention.

JUDGE CRISTAUDO: Thank you.

COMMISSIONER ASTRUE: I hate to cut this off because it's very helpful and -- and really interesting, but it's my sad duty to do that.

Thank you to all the panelists. It's been very helpful.

I want to reiterate our gratitude to Dr. Koroshetz and -- and all of NIH. They've just been great partners on this, and I think they're going to be incredibly important for us going forward, and we're -- we're very grateful for the support.

We're going to take a 15-minute break

now.

I have concluded that we were overly generous with the closing remarks for the panelists here, and I don't think we're going to use that time. So we'll reconvene at 3:20. We'll have an hour for the final panel.

And then we'll -- we'll -- we'll restrict ourselves to no more than 10 minutes. I'll still get you out of here by about 4:30.

So thank you very much, and we'll reconvene in 15 minutes.

(Recess.)

COMMISSIONER ASTRUE: Okay. We're on the home stretch here. We've got one last panel, which I'm looking forward to, so I -- I'd like you to interrupt the conversations again and focus our attention.

We have four members of this panel. We have Dr. R. Rodney Howell, who is President of the American College of Medical Genetics Foundation and Chair -- and he's also chair of Secretary Leavitt's Advisory Committee on

Heritable Disorders and Genetic Diseases in Newborns and Children.

We have Dr. William A. Gahl, who's clinical director of the National Human Genome Research Institute at the National Institutes of Health with his colleague, Dr. Suzanne Hart.

And finally, we also have Dr. Andrea Gropman, Associate Professor of Pediatrics and Neurology at George Washington University School of Medicine and Health Sciences, attending in neurology, who's also with the Children's National Medical Center.

Welcome.

Who -- who's going start? Dr. Howell, are you going to start?

DR. HOWELL: I think that's the order.

COMMISSIONER ASTRUE: Okay. Great.

DR. HOWELL: Thank you very much, Commissioner Astrue. It's my pleasure to be here. And I'm going to start with some very general overview comments about newborn screening.

The newborn screening has been an

active program in the United States for a little over 40 years. It started with the recognition that a long-known condition, that is, phenylketonuria, an abnormality in the metabolism of phenylalanine, could be dramatically improved if infants, very early in life, were put on a special diet that was deficient in phenylalanine.

The infants who were treated early were recognized because they had a family history of the condition so that they could be tested when they were little babies and so forth and tried on the diet.

It was also shown very early that if you tried this new diet on older children with phenylketonuria, the benefit was minimum. So that you had the very striking paradigm, a dramatic diet that had to be diagnosed in the newborn period. And since there's no family history usually in these conditions, you needed to conceive of a test that could be applied to the population at large.

At that time, the -- the question was

posed to Bob Guthrie, who is a Professor of Pediatrics and Microbiology in Buffalo, who was active in the community of persons with handicapped, about could he devise a test. And he come up with a very simple bacteriologic assay that depended on using a dried blood spot on filter paper and putting those dried blood spots onto auger containing a B subtilis spore that was dependent on phenylalanine. And this very simple test turned out to be extremely usable in the public health arena.

And largely because of public advocacy, families with children with phenylketonuria, this technology was adopted rapidly throughout the country over a period of several years, and this was in the early to mid '60s.

The -- the situation stayed relatively the same, technologically; that is, a dried blood spot, you punch out one hole, and you test for one condition. However, other conditions were added that had therapies, such as maple syrup urine

disease, hypothyroidism, but you had this one spot, one test technology. Some states actually ran as many as 10 tests, all on individual little holes that they punched out as tests.

The scene changed quite dramatically in the early '90s with the widespread adoption of tandem mass spectroscopy. And this technology is extremely reliable and permits you, on a single dried blood spot, to analyze for a number of abnormal compounds, and in the areas of inherited rare metabolic disease, it turns out that you can really diagnose around 40 conditions in -- in a matter of minutes with considerable accuracy.

And since that time, there's been a considerable increase in the number of tests that are done in newborn infants in this country. At the current time, the national recommendations from both the advisory committee that I chair and also from the American College of Medical Genetics is that there are 29 conditions that are appropriate for newborn screening in the so-called core panel.

These conditions all share the common denominators: One, they're all very serious, they all have treatments that, if begun earlier, are beneficial. And the other thing is they're not evident at birth; in other words, the baby looks normal, so there's no sign that would say, goodness, this baby has this problem.

Now, state newborn screening -- newborn screening programs are, in all instances, operated by the states. And so what is screened in Virginia, is decided in Virginia, et cetera. But it's very been interesting to me from a national level to look at what's happened since these recommendations have gone down in the past few years. Because as we sit here today, 90 percent of all the babies born in the United States are getting that -- that core panel done. And so the states have actually quite rapidly adopted that panel.

It'd be appropriate to comment at this time is that there are 4.1 million babies born each year in the United States, and since they are

all getting genetic testing and many or most of them, as many as 30 conditions, newborn screening for genetic diseases is by far the most common use of genetic testing that's done today in this country.

I think these conditions are particularly interesting for a variety of reasons. One is that people with these conditions are born with what I would call a permanent abnormality or disability. In other words, they lack a protein or they lack an enzyme. However, if detected very early, they can commonly lead an essentially normal life.

And recent -- the recent NIH panel on the treatment of phenylketonuria had a college student as a member of the panel who herself had PKU. An untreated PKU, the average intelligence is under 25, so the benefits are enormous.

However, there are many programs currently being screened for in the country that are not, shall I say, "curative." For example, most states currently screen for cystic fibrosis

and although the outcomes are improved, children with cystic fibrosis persist with disability, as you know, although they do much better if diagnosed early, et cetera.

The other widespread screening condition today that falls in a similar category is all states are either implementing or currently screening for hearing defects.

And although you can benefit these children by early identification of the defect, they commonly will need either cochlear implantation or assistive devices in order to hear. So they will need long-term support and help.

And I'd like to just comment about the fact that there are dramatic new technologies on the scene. I've talked about tandem mass spec and so forth. But currently under look -- under investigation are some microfluidics which use extraordinarily small samples and can screen for a large number of -- of areas.

There are new diseases that are being

screened for that have therapies coming up. Two lysosomal storage diseases that are currently in pilot studies in the United States for newborn screening are Krabbe disease, a historically rapidly fatal disease that, if identified in early infancy and treated with a bone marrow transplant, has dramatic improvement. These children are not cured, they still have disability, but they're dramatically improved and they survive.

The other is Pompe disease, a lysosomal storage disease that, in its most severe form, causes early infant death, is under treatment with enzyme replacement therapy, which is being introduced for a number of conditions, as the Commissioner certainly knows, having been seminally involved in this area himself in the past.

I think that the -- the -- the point is, is that the technology that's being used in newborn screening is highly accurate. One commonly talks about, you know, you have a certain number of false positives, and I would say that

the term "false positive" is not the right term, because false positive suggests that you have an error in laboratory diagnosis or you have something, but we have physiologic variance with chemical changes in the baby. And so a test may not be within your normal limit and when repeated, is normal.

But I would call we need to come up with a better term. There's variation and so forth, but the technology that's being used is extremely reliable and very accurate.

And I think one of the major things that's come out of the newborn screening needs in recent years has been the fact that once we identify an infant -- and about 1 in 1,000 babies born in this country will screen positive for one of these conditions, so it's a substantial number of -- of infants. Although they're individually rare when you put them together, it's a substantial number.

We need to develop a system to put all of those into a long-term follow-up program to see

exactly how they're doing with treatment, what's happening to them, and also make available these children and young adults for new -- new therapies and new technology.

And this is one of the areas where the National Institutes of Health is looking very carefully at how to institute -- is to implement a program that would study the -- all the babies that are born positive in this country.

So those are my opening remarks. Thank you very much.

COMMISSIONER ASTRUE: As has become traditional today, we're going to hold off our questions until the end and then -- because we figure there'll be overlap in response. Okay?

DR. GAHL: Okay. Thank you.

Well, medical technology can bring us to a diagnosis and can often bring us objectivity to that diagnosis, and sometimes the diagnosis can bring us a prognosis as well.

So Dr. Hart is going to talk about some specifics of those medical technologies, and

then I'm going to offer a perspective after -- after she speaks for just a few moments. And Dr. Hart has wide experience in molecular and biochemical diagnostics, so she's going to tell us a few things about those specifics.

DR. HART: The Task Force on Genetic Testing, convened by the National Institutes of Health, Department of Energy Joint Working Group on the Ethical, Legal and Social Implications of Human Genetic Research, defined a genetic test as:

The analysis of human DNA, RNA, chromosomes, proteins and certain metabolites in order to detect heritable disease-related genotypes, mutations, phenotypes or karyotypes for clinical purposes. Such purposes include predicting risk of disease, identifying carriers, establishing prenatal and clinical diagnosis or prognosis. Prenatal, newborn and carrier screening, as well as testing in high-risk families are included.

Tests for metabolites are covered only when they are undertaken with high probability

that an excess or a deficiency of the metabolite indicates the presence of heritable mutations in single genes.

Tests conducted purely for research are excluded from the definition, as are tests for somatic, as opposed to heritable mutations and testing for forensic purposes.

Individuals who undergo genetic testing fall into two main categories: One, symptomatic individuals are those in whom a specific clinical feature or phenotype suggests a genetic disorder; or, two, individuals with a family history of a genetic condition.

Current genetic testing methodologies are broadly categorized into biochemical, molecular and cytogenetic techniques.

This testimony will focus on biochemical and molecular techniques.

Existing molecular genetic testing includes targeted mutation analysis, for example, by targeted sequencing, allele specific oligonucleotide analysis, et cetera, whole gene

sequencing, typically covering coding regions and exon/intron splice junctions, determination of number of repeats for trinucleotide repeat disorders, deletion/duplication analysis, and determination of epigenetic changes such as methylation status.

Detection rates vary based upon the gene and the methodology used. As sequencing costs have fallen, full sequencing has become more common. This will likely result in higher detection rates compared to targeted analysis.

In the future, array technology will be more frequently employed in the molecular diagnostic laboratory. There are two main areas where this technology will be helpful. First, for large genes, array technology provides a cost-effective and time-saving platform for mutation detection.

For example, the dystrophin gene, which is mutated in the two allelic disorders, Duchenne muscular dystrophy, DMD, and Becker muscular dystrophy, spans more than 2 million

basepairs of DNA.

More than 500 deletions, 80 duplications and almost 1,000 single base changes have been described in this gene. Two arrays have been developed to aid in genetic testing for these muscular dystrophies. The first array detects deletions and duplications of the DMD gene, which account for approximately 70 to 75 percent of mutations in DMD and 90 to 95 percent of mutations in Becker muscular dystrophy.

The second array is a sequencing array that allows the whole DMD gene to be interrogated in a single assay.

Another use of array technology will be the evaluation of multiple genes simultaneously. The advantage of this technology can be illustrated with X-linked mental retardation.

Currently, 63 genes on the X chromosome are known to be associated with syndromic X-linked mental retardation. This is

mental retardation associated with other phenotypic features.

An array is under development that would allow analysis of 34 genes on the X chromosome concurrently. At the present time, such analysis would have to be conducted sequentially at a significantly higher cost and turnaround time.

Biochemical testing for rare diseases involves standard blood chemistry, hematological and immunological studies, as well as specialized tests for rare metabolic disorders. These include amino acid and organic acid analysis, enzyme assays, antibody tests, specific small molecule assays and profiles of abnormal metabolites.

As Dr. Howell has mentioned, tandem mass spectrometry is revolutionizing newborn screening, and this method has the potential to improve the biochemical diagnosis of symptomatic patients as well as newborns.

The technique can allow faster and perhaps more definitive diagnosis of known

diseases and can reveal new disorders by detecting small molecules that signal disease.

Ultimately, the best testing methodology varies with the disorder or gene involved and may also depend upon ethnicity. For example, carrier testing for Tay Sachs disease, in the absence of family history, is best performed by molecular means for Ashkenazi Jewish individuals, but by biochemical methods for nonAshkenazi Jewish individuals.

In some cases, combined testing may be the most appropriate approach.

DR. GAHL: I would say that these medical technologies get us to a diagnosis, and sometimes that diagnosis is incredibly meaningful in itself; that is to say, that everyone who has the diagnosis will have a disability. And examples of that could be Progeria, which is a premature aging syndrome in which the patients die at about 13 years of age. Or a Fragile X or Tay Sachs disease, which is a lysosomal storage disorder, or other lysosomal storage disorders,

adrenoleukodystrophy, for example, Usher's syndrome, which will predict deafness, or albinism, which will predict blindness.

But then there are other disorders in which the diagnosis does not itself predict disability, and you need to know something else about it. For example, in Huntington's disease, you know eventually there's going to be problems, but you may not -- if you carry the gene, you may not have the problem at age 15.

And the same goes for some of the late onset myopathies like Hereditary Inclusion Body Myopathy and even Fabry disease, which has later onset to these disorders.

Lists of these types of disorders, the ones that by themselves are going to give a problem immediately and the ones that may later can be constructed with some reasonable objectivity because of medical technology and expertise. And that's the second point that I'll come to in a little bit.

But most patients who come with a rare

disease diagnosis will eventually require medical assistance. The issue is at what time is the diagnosis being made, what time are they being evaluated. If they come to medical diagnosis and use medical technology at the time when they have symptoms, they probably already need intervention.

But sometimes the genetic diagnosis is made before that, and it's only later that they'll need to -- to be declared disabled and -- and receive assistance.

Furthermore, there are different rates of progression of these disorders, and some of the disorders are even static.

Well, I think that the new technologies that will come about will not change this basic paradigm incredibly much. In other words, the diagnosis will always take us so far, but never so that we can completely predict for all of these rare diseases that disability is required.

The bulk of the decisions regarding these allowances also require input from health

care professionals, experts in the field, and fortunately, for many of the rarer diseases, there's a -- for the metabolic disorders, there's a society for inherited metabolic disorders, and that society has those experts. And I think that as a group, the SIMD would be willing to offer its expertise in the adjudicatory process, along with other health care professionals.

So they could do this on an individual label -- basis, but they could also offer advice a priori by creating some of those lists of the disorders which are certain to require disability allowance and those that may or may not, based upon particular symptoms.

Thank you.

DR. GROPMAN: First I'd like to say it's my pleasure to be here today, and I commend the Commissioner and his committee for convening this hearing.

As you've heard from my colleagues, our knowledge of inborn errors of metabolism and our screening in recent years has expanded at an

impressive rate as a result of research in laboratories and clinic -- clinical trials around the world.

Rapid diagnosis and, when possible in treatable conditions, metabolic rescue, is mandatory to improve neurologic outcome as the majority of these conditions that were discussed impair the neural access in some way.

In addition, advances in technology have made early diagnosis feasible, and with this recognition, the outcomes of treatment better.

As new treatments are emerging, they will impact on outcomes. One consequence that you've heard of early screening and treatment is that children with these diseases that were once considered uniformly fatal are now surviving into adulthood; some as healthy productive citizens, others with significant challenges ahead of them.

Despite these advances, however, there continue to be barriers that exist leading to diagnosis and provision of services to individuals with these conditions.

Many of these conditions are expected to worsen over time. And although physical disability may be expected, the impact on neurologic function, cognitive changes and mental function may be less clear.

Likewise, the discovery of new variants of previously described metabolic diseases has added further to the complexity of diagnosis of inherited metabolic disorders.

Late onset in adult variants of diseases formerly thought to occur only in infancy or childhood are being diagnosed not infrequently. And often, the clinical features of the disease in older individuals is markedly different from that seen in infancy.

As a child neurologist and geneticist, I have firsthand experience in the problems of getting to diagnosis and being able to determine disability.

Neuroimaging of inborn errors of metabolism has begun to paralyze advances in other technologies. As the central nervous system

effects of some congenital and acquired infections are recognized by characteristic distributions of abnormalities on neuroimaging studies, likewise, specific imaging findings may, in some cases, strongly suggest a metabolic disorder or inborn error of metabolism as the etiology of the patient's neurologic symptoms or disability.

In fact, large collaborative research studies, predominantly in adult degenerative disorders, have convened to look for biomarkers that may actually predict the onset severity as well as response to treatment in a number of conditions which could impact the determination for disability.

While in other cases the imaging features of metabolic disorders may mimic that due to other causes, there are certain well-described imaging patterns that strongly support the diagnosis of a metabolic disorder.

These patterns, in turn, may depend upon the age and stage of the disease at the onset of symptoms, and furthermore, cell type and

regional selective vulnerability to metabolic toxins and ischemia are responsible for the differing imaging patterns that are seen.

The differential diagnosis of a metabolic disorder is thus narrowed, depending on whether the findings affect primarily the gray or white matter, both, and there is typically an imaging signature that may be recognized, such as an altered signal in the basal ganglia in glutaric aciduria Type I, the parietal occipital changes seen in MELAS and other mitochondrial disorders, absence of the creatine peak on magnetic resonance imaging spectroscopy seen in creatine deficiency disorders, or an increase in N-acetylaspartate, a putative neuronal marker which is seen in Canavan's disease.

Likewise, a common imaging finding in patients with changes in mental status may be that of edema caused by vasogenic or cell swelling and may accompany stroke, infection or metabolic disorder.

The location of the swelling, for

example, being in white matter, brainstem and cerebellum, would invoke the diagnosis of maple syrup urine disease, for example, and not hypoxic ischemic injury in an infant presenting with changes in mental status and edema.

Certain organic acidemias, such as propionic or methylmalonic have a predilection for causing signal changes on MRI in white matter and basal ganglia, and the urea cycles classically show elevations of glutamine, as can be measured noninvasively by magnetic resonance spectroscopy.

Because inborn errors in metabolism can present in the neonatal period with neurologic distress, metabolic acidosis and multisystem involvement, similar features encountered in hypoxic-ischemic encephalopathy, an underlying metabolic disorder may be undiagnosed, unless the clinician maintains a high index of suspicion and performs appropriate diagnostic testing.

This certainly will impact the ability to recognize and provide needed services for these individuals.

So, as I've indicated, magnetic resonance imaging has emerged as a powerful tool in the study of both normal and abnormal brain structure, function and biochemistry.

It is a nonionizing and noninvasive technique and most, if not all, medical centers have access to one or several scanners, and imaging of pediatric patients, including our premature infants, is now quite routine.

While skillful assessment of the results is required, there are several references to assist with this process. MRI can allow the differentiation of cell bodies or gray matter from the connecting fibers, or the axons, which appear in the white matter.

And with a technique called magnetic resonance spectroscopy, or MRS, brain biochemistry can be sampled in a noninvasive way, and this information taken with the clinical features to aid with diagnosis.

In addition, newer modalities of imaging are moving from the research scanners into

more widespread clinical use. For example, white matter microstructure can be studied using a technique called diffusion tensor imaging, which may allow abnormal white matter to be visualized prior to seeing it on conventional MRI.

And magnetic resonance spectroscopy, to study brain metabolism, can allow for delineation of regional metabolic changes as a result of disease progression or, as therapeutic interventions are introduced, can be used to measure outcome.

Neuroimaging studies may be indicated in a majority of patients with mental retardation, developmental delay, acute encephalopathy, loss of skills or cognitive impairments of unknown etiology.

Even in conditions that are due to known causes, the MRI can be used to assess the degree of involvement, which may help contribute to disability determination.

The most common imaging techniques used currently in the diagnostic evaluation of

individuals with mental retardation or disabilities are CAT scan, MRI and magnetic resonance spectroscopy. However, given the unique gray/white matter differentiation, MRI is probably the best imaging test in a diagnostic workup.

Some conditions in which neuroimaging procedures, and specifically MRI, have proven to be useful include the following cases, which I'll discuss:

Neuronal migration disorders, brain malformations or hypoplasias, intrauterine infections, disorders due to birth injury, vascular disorders, white matter diseases, including demyelinating disorders such as ALD, multiple sclerosis, metachromatic leukodystrophy, and Canavan disease; and other metabolic conditions, organic acidemias, mitochondrial disorders, urea cycle disorders.

Because of the huge array of available molecular and biochemical tests, the information from neuroimaging may help focus the search.

The technologies of today that are

useful within the MRI scan package itself include FLAIR imaging to view white matter, MR spectroscopy and diffusion tensor imaging.

And in the future, improvements of these methods with higher field scanners and other types of metabolic imaging will certainly improve early diagnosis.

Therefore, the delivery of SSI and SSA services to individuals with rare disorders will depend upon early diagnosis, understanding of the diagnosis -- of the disorders and their effects on function and independent living, and understanding the limitations may not be static, but, in some conditions, are expected to be progressive over time.

The major challenge and question, therefore, is to how to use and integrate these technologies in a way that will impact disability determination with the goal to establish an accurate and valid system that benefits those patients in needs of services.

I believe I speak also on behalf of my

colleagues that, as a group, neurologists and geneticists are available to engage in such dialogue to address this, as we appreciate the importance and complexity of this disability determination process.

Thank you.

COMMISSIONER ASTRUE: Thank you. Thank you. I've got a couple questions.

We, of course, because of our mission, have to ask very different questions than what doctors trying to treat disease or companies trying to provide therapies ask.

So we're -- we're very focused on trying to figure out how markers correlate with functionality.

So I wanted to ask first on -- on genetic testing. A lot of the lysosomal storage diseases, for instance, have a wide range of severity. Pompe disease, you can have children dead at the age of 2, but I've also had the experience of talking to a woman in her 30s for 15 minutes before she disclosed she had

Pompe disease, and I had no clue.

So there -- there's a range, and something like Hunter's, somewhere, 5, 6, it'll split pretty evenly between those kids that have a pretty good shot at not only survival, but living pretty normal lives, with some of the new therapies and those where the disease attacks the central nervous system, and they're looking at a horrible death, you know, in a five- to eight-year period.

To what extent are we able now, through genetic testing, to sort out within those diseases that have a range of severity, can we tell genetically at birth what the likely course is? For instance, with a Hunter's child, which one is likely to have seen as complications or not or -- or is that we're just not there yet in terms of being able to use this -- these data?

DR. GAHL: I'm -- I'm going to let Andrea answer in a minute, but I don't think we're really there yet with respect to genetic testing. It turns out that there's relatively poor, or

let's say incomplete genotype, phenotype correlations for most of these disorders. And that may fill out over the next, let's say, decade or two, because the clinical characteristics are then associated with certain mutations.

But there's a problem here, maybe not quite so much for Hunter's as for some of the others that are autosomal recessive, you have compound heterozygotes -- so, in other words, two different mutations -- and you can't really tell -- you can tell that there's a severity associated with one mutation, but not so well when there are two different mutations, essentially. So -- so, you know -- so -- so that's -- that's an issue.

It turns out that some of our best phenotype correlations are with biochemical measurements. So the amount of residual enzyme activity does correlate pretty well with the severity of disorders, and that goes way back.

So we should perhaps move ourselves a little bit away from the molecular and towards the

biochemical, which essentially measures all sins. You know, it's the final measure of how much residual competency you have for that enzyme pathway.

I'd also say that differential tissue expression can be a problem here. In other words, you might have a particular mutation in a gene such that there's a reasonably normal amount of enzyme activity within the liver or the kidney or some such, but not so much in the brain, and that can account for some of -- some of the differences, too. And we're not very good at that, because we don't get those organ or tissues to measure the enzyme in.

COMMISSIONER ASTRUE: What -- what percentage of diseases? Is it just a question of brute force as opposed to sort of an intellectual issue that needs to be resolved? Do you have any sense of that?

DR. GAHL: What -- what percentage of disease is what?

COMMISSIONER ASTRUE: Would -- would

we be able to perhaps sort out the severity of the disease from gene testing as opposed to waiting to see what happens? Is it -- is -- is it possible, do you think, in 5 percent, 10 percent if we do the work? Is it higher than that?

DR. GAHL: I -- I think it's higher than that, actually. It -- it has to do with accrual of patients and performing clinical characterization at the same time as doing the molecular diagnostics. So to a certain extent, this is a matter of time and -- and money.

You could probably do it now for half or more of the disorders. It's just that, you know, there aren't grants for that, there's no real support for that.

COMMISSIONER ASTRUE: Right.

DR. GAHL: Companies are coming close on occasion to doing that because it's sort of in their best interests.

COMMISSIONER ASTRUE: Okay.

DR. HOWELL: Let me comment. I certainly agree with everything that Bill said and

the -- I mentioned the fact that the NIH is very interested in following up on the newborn kids, and one of the things that will be looked at keenly is looking at all the markers, looking at enzyme activity and correlating that with long-term follow-up and see how that works out.

Now, the other thing is that New York State has had to deal with this in spades in their screening program for Krabbe disease. They have mandated screening in the state for Krabbe disease and, like Pompe disease, there's different forms of Krabbe. Some are acutely ill as infants and others have a later onset.

And so what they've done, and I think they worked very hard on this, is to, once the patient is identified as having an enzyme deficiency, they basically then have a whole variety of other tests that they put into play, such as nerve conduction velocity, there's a variety of other things, and try to make a judgment about, you know, what's going to be the time frame of that patient. And I think those

will clearly be informative when -- when they -- when they finish their studies over this time.

But they've had to deal with -- and Pompe, of course, is a perfect example. The -- Bill mentioned the professional groups are looking at paradigms in the diagnosis and treatment and the -- the college has been working with SIMD on a paradigm in the diagnostic follow-up of patients with Pompe disease. Because once you get a patient with Pompe disease, you're going to have to make some decisions fairly early, because some of the patients will have massive cardiomegaly and others will be asymptomatic.

And I think doing genotype analyses will be helpful, but the correlations have not been very good. But enzyme activity, according to the Duke group, has been fairly predictive.

COMMISSIONER ASTRUE: Okay. I've got one more question, and then I'll turn it over to my colleagues. I'll start with Dr. Gropman, but I don't, you know, want to stop anyone else from jumping in.

And this is a little bit out of our mandate today, but that's okay. You're all here, so I'm just going to ask what I want, so --

We -- we have -- one of the -- we have a lot of claimants in certain categories in the neurological diseases, and it can often be very hard for us to sort out for these degenerative diseases sort of the break point where they fit the standards that Congress has told us that we -- we have to apply.

And so one of the things we first started to talk to NIH about was multiple sclerosis and looking at whether we could look at a measure like neuronal scarring in the brain and correlate that with functionality.

And one of the things that came back at -- at some point was, well, the brain as a whole, that might be difficult, because depending on -- on where the scarring is, that might make a big difference in terms of functionality.

Do you have any advice to us if we're -- we're -- I mean, one -- one of the things

that would be a blessing for us and for -- for claimants if we could sort it out would be some efficient ways to handle these types of neurological diseases.

Do you think that we're likely to find certain areas of the brain, if we focused on sort of subsets, not the brain as a whole, and could track damage like neuronal scarring in MS, are there certain areas of the brain that perhaps we ought to be more focused on and we're more likely to be able to prove some sort of correlation to functionality?

DR. GROPMAN: I think it's more complex than that. And basically --

COMMISSIONER ASTRUE: I was afraid of that.

DR. GROPMAN: Yeah. Unfortunately, it has to be on a case-by-case basis, because with some of these metabolic conditions, I didn't mention that in addition to the enzyme defect, there may be other compounding factors, such as a patient may have seizures, for example, which adds

further disability.

You know, it -- certainly the cortex being affected will impair cognitive function, but that's not to say that having a lesion in the brain stem which affects basic functions such as swallowing and -- and the like isn't significant as well.

So it -- and, you know, certainly a cerebellar lesion can affect the ability to walk, you know, without falling.

So I think, you know, it will have to be a case-by-case basis. I -- I believe that we do have limitations with imaging.

We certainly have a number of impaired children who have, at least structurally, normal MRI scans, so we know that there's more to be told than just, you know, the brain looks fine, but obviously it's not functioning. Well, we're not able to image on the level of synapses, for example, in kids with autism and, you know, we're not there yet.

So I think it's more complex than

that. You may -- for each disease group, may be able to categorize disability some, but I think just taken in general, it's much more complex than that.

COMMISSIONER ASTRUE: Okay.

DR. GROFT: A -- a couple questions. I guess first, how -- how many inherited metabolic disorders would fall under the umbrella of the SIMD, the group that would look at those diseases? Do we have any -- any approximate numbers?

DR. GAHL: Well, Andrea, you -- you can help me out on this, but probably in the range of 100 or so. There are 225 or 250 members -- well, actually, there are 450 members of the society, but in terms of actual physicians taking care of patients, there are probably about 225 or so. And they overlap some. So I'd say maybe 100, 150 different diseases.

DR. GROFT: And -- and so the society members that you have probably would have pretty good knowledge of most of these disorders that -- that they really could -- you could bring them

together without too much difficulty to -- to address the issue of, you know, the level of functioning and --

DR. GAHL: Yes. I think the SIMD recognizes its service role to the community and to the country. And I think that they would be anxious to offer information on this and to assist in something that the Social Security Administration wants -- wants to establish.

Basically, the physicians in the SIMD take care of these patients and deal with these issues all the time. They actually write letters for their patients to the Social Security Administration.

So they're very interested in the -- in the care of -- of the individuals with metabolic diseases. And there's a great lack of understanding on the parts of -- of people all over the country concerning the severity and the functional effects of these rare disorders.

DR. GROFT: Okay. Thanks.

ACTING DEPUTY COMMISSIONER RUST: Sort

of following up on the line of questioning the Commissioner started with and also the last panel, they began the discussion to -- or surfaced the idea that there are scales that researchers and physicians use to measure severity in many of these disease categories.

That -- that interests me a great deal because one of the things we would like to do is begin to find more objective measures of the -- the severity of -- and the disabling aspects of -- of disease.

Can you comment on that or add anything to that discussion?

DR. GROPMAN: Okay. I think there are some limitations with the scales, one being certainly many of them don't apply to pediatric neurologic patients. I'll take MS as an example.

So MS in children can be very different than it is in adults, and those scales may not apply. Even just to take diagnostic scales, the criteria for NF1, which, you know, were ingrained in all of us going through medical

school and training, really, you're hard-pressed to make a diagnosis in a child under, you know, puberty age using just, you know, the -- the diagnostic criteria from the NF1 collaborative scales.

So I think -- and -- and child neurology society is keenly aware of the fact that we don't have scales to the level that our adult neurologic colleagues do and are actively working on it.

I know, you know, for cerebral palsy, there are some -- there are scales. There's a book called, you know, Book of Scales, but whether they're all applicable, you know, is another issue. So I think -- and the same would go for some of these metabolic conditions, because even children with the same metabolic disorder may present different manifestations. One child may be still ambulatory with predominantly cognitive effects, whereas another child with the same disorder may be in a wheelchair, for example. So --

COMMISSIONER ASTRUE: If -- if I could cut in, I don't want discourage, but I just have -- want to ask a specific question. For the childhood MS cases, are those always going to end up being severe cases as adults, or do you get cases where you have onset very early and it's very intermittent, the way sometimes some of the adult cases are?

DR. GROPMAN: I think the story's just unraveling with childhood MS. You know, certainly making the diagnose -- the definitive diagnosis of MS in a child versus its close cousin, or acute demyelinating encephalomyelopathy is -- is a problem. You know, a child who presents with one demyelinating event, you know, what is it about their event or them that will predict whether they have MS or -- or they don't have MS.

So I think that's -- that's unraveling now. There is a working group within child neurology society addressing those issues.

DR. GAHL: I'd like to sort of interject, too, about the point about geneticists.

We see patients who often have multisystemic disorders, so that there isn't really so much a scale with respect to one particular symptom, it's a question of whether, for example, a cystinosis patients -- patient has just renal disease or also has muscle disease, swallowing difficulties, pulmonary disease or central nervous system problems or diabetes because those are complications, and they're fairly discrete as opposed to scaled. You know, they've got it as a complication or not.

When I need functional scales, I generally get it from the rehabilitation medicine folks, who are very good at -- at this and -- and measure function in terms of range of motion or abilities to do certain activities of daily living, and they're the best for me when I need a consultation.

COMMISSIONER ASTRUE: Frank.

JUDGE CRISTAUDO: I would like to ask the question that I've asked the earlier Panels. Obviously a very distinguished panel, and it would

be very helpful.

Obviously, it's critical for us to have the relevant information about the individuals' impairments and so on.

Is there anything that we could be doing in -- in terms of when we ask for information from medical sources to make it easiest for the medical sources to provide the information to us?

If anyone has any comments on that.

DR. HOWELL: I don't have any comments on it.

DR. GAHL: Are -- are you -- are you sort of asking about Freedom of Information or are you asking for specific information to be garnered?

JUDGE CRISTAUDO: See, we're -- we're trying to make determinations whether or not the claimants' impairments meet a particular list of impairment in our -- in our listing of impairments, or we're trying to assess function, and some of the earlier questions have referenced

that.

So the question really is, is we have some difficulty sometimes having medical sources provide information to us about -- about their patients, essentially. So what -- what I'm looking for is, is there an easier way for -- for medical sources to provide that information to us? Should we be asking the questions differently? Is there anything else we can do to facilitate that process?

DR. HOWELL: I think Bill has made an important comment, is that the spectrum of conditions we work with is such -- for example, in Pompe disease, you need a cardiology evaluation, the cardiac problem, et cetera. Another, you have, in some of the others, you may have a neurologic problem completely, you have a neuromuscular.

So I think a task force, shall we say, at one of the referral centers is I think the thing that's going to be helpful, and -- and you're going to need all of that.

DR. GAHL: I -- I think -- I may -- I think --

COMMISSIONER ASTRUE: Not to jump on the -- the Panel's answer. I mean, I think one of the things that's going to be important for us as we start listing the -- these diseases is to flag for the examiners the type of tests they should be running, because they don't know, because they don't have familiarity with the disease.

You know, they could see a description of Fabry disease and not realize that you should be looking primarily probably at the heart and the kidney. And it will -- could be an awful lot more efficient for us if -- if we have instructions to our examiners where, you know, that's sort of flagged out for them instead of trying to get them, their general doc to find an expert who can give them that answer, and a lot of times we have a lot of difficulty getting expert consultants in these -- in the rare disease areas.

DR. HOWELL: One of the things I think can be very helpful along that line are the

practice guidelines that are being developed for virtually all of these conditions that basically will flag, you know, what other systems that become involved. And they currently are in the process of being developed for many of these. They've been developed for Pompe and Fabry and Krabbe, but they're coming along for the others.

COMMISSIONER ASTRUE: And who generally has the lead in those practices?

DR. HOWELL: The American College of Medical Genetics. Again, a lot of overlap with SIMD because we have -- I'm a former president of SIMD, but I currently am now with ACMG, so there's a lot of overlap between the groups, but they work together on those. They're usually published under ACMG Guidelines.

DR. GAHL: And I do think that for a particular diagnosis, you could probably, a priori, get SIMD members who are experts in that to create a list for that particular disease for -- for -- for you to request from the physician. And you could probably do it for many,

many disorders; not complete, but -- but many, and that could serve as sort of the first measure or guide for what you need to make that assessment. And I think the SIMD and other experts would be willing to do that.

COMMISSIONER ASTRUE: Thank you. Go ahead.

ACTING DEPUTY COMMISSIONER RUST: If I can sort of take another tact on -- on Judge Cristaudo's question.

Our examiners are looking -- the -- the treating physician is looking at the patient one way, and our examiners are looking at the patient another way. And we sometimes have a sense that we don't ask for the right material or the -- the physician doesn't send us the material we need to -- to -- that there's a -- there's a gap in terms of understanding or a -- a mindset between the -- the treating physician and the examiner.

And if we could bridge that, we might get the right answers to our questions that would

help us make that disability determination. I -- I think that's what we've been wrestling with, how do we get that.

DR. GAHL: Well, I -- I could just sort of reiterate what I said about function. In other words, you're right, physicians don't think about function very often. They're happy to make a diagnosis and give some medications, et cetera.

But the -- the physical therapy and the rehab folks do think about function all the time. So perhaps you could require that there be a rehab medicine type consultation on this, you know, what can the person do and what can't the person do attendant to this diagnosis.

What do you think about that?

DR. GROPMAN: Yeah. Yeah, that makes sense. Yeah. I think, you know, neurologists are focused somewhat on function, but certainly our rehab colleagues, that's the bane of their practice, and to say, okay, you know, thank you, we have the information about the diagnosis, we need to determine the functional limitations of

this individual.

I would think that perhaps some places would not necessarily have access to physical medicine, so basically laying out exactly what a functional determination is might be helpful in those cases.

COMMISSIONER ASTRUE: Do you have anything else?

JUDGE CRISTAUDO: No.

COMMISSIONER ASTRUE: Do you want to make any final remarks?

ACTING DEPUTY COMMISSIONER RUST: I -- I would just like to -- to thank all the witnesses today, all the Panel Members, because the information has been very helpful, and I think it's going to -- going to go a long way to helping us tackle this particular problem.

COMMISSIONER ASTRUE: I know, I've just checked with Steve and Frank, they don't have any final remarks except also to thank the panelists and everybody that's participated today.

I just want to say it's been -- this

has been extraordinarily useful. Just to follow up and on everything that's been said today that's helpful, it's going to take a very, very long time.

But I do want to remind people in the audience that, from our vantage point, this is not a one-shot exercise, and that it's our intention to have these types of hearings on a quarterly basis on a different -- different subject each time.

And we have blocked out a schedule for the rest of the year and have the next three meetings set, and as we get into the summer, we'll have the next year meetings set. And by the time we've exhausted all the obvious things, there will be time to go back and do updates on technologies on the others.

So this is -- from our vantage point, we're trying to fundamentally change how we access medical information and -- and as you can see from today, there's so much that's out there, it's so hard, it's so complicated, that if we just try to

do it in our own closed world, as wonderful and talented as our doctors are, we can't possibly do it all. And so we're trying to find more ways to reach out and get as much help from experts in the field as possible.

I think this is a terrific way to start. I think all four of these Panels were extremely helpful to us. And so I want to thank everybody who participated, Dave and Diane and Glenn and the Staff that put together this terrific program. And thank you all, and I -- and I'll let you out 17 minutes early.

So, again, thank you all.

(Applause.)

(Whereupon, the Outreach Hearing adjourned at 4:13 p.m.)

CERTIFICATE OF REPORTER

I, CINDY L. SEBO, Court Reporter, do hereby certify that the testimony that appears in the foregoing transcript is the testimony of said witnesses, were taken by me in shorthand and thereafter reduced to computerized transcription under my direction; that said transcript is a true record of the testimony given by said witnesses; do hereby certify that the foregoing transcript is a true and correct record of the statements of counsel; that I am neither counsel for, related to, nor am employed by any of the parties to the action; and further, that I am not a relative or employee of any attorney or counsel employed by the parties thereto, nor financially or otherwise interested in the outcome of the action.

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