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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

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

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

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

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

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

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

1 here, and we appreciate their hospitality.

2 MS. BRAXTON: Thank you.

3 COMMISSIONER ASTRUE: Good morning.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

10 Can you hear me in the back, anyway?

11 UNIDENTIFIABLE VOICE: Yes.

12 COMMISSIONER ASTRUE: Yes.

13 Okay. Good.

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

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

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

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

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

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

17 UNIDENTIFIABLE VOICE: Excuse me. Is
18 someone on line?

19 MS. WILSON: Yes.

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

1 is saying.

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

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

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

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

17 MS. WILSON: Yes.

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

20 MS. WILSON: Thank you.

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

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

5 (Laughter.)

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

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

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

22 These are precisely the types of cases

1 where examiners are uncertain as to what to do.

2 This uncertainty leads to requests for

3 time-consuming and expensive consultations often

4 performed by doctors that are unfamiliar with our

5 rules and regulations.

6 In particular, this is why our first

7 hearing focuses on rare diseases. If the process

8 can be approved for children and adults with rare

9 diseases, it can be approved for everyone.

10 Second, we need to fast track the

11 cases that are certain or near certain to be

12 allowed, and with our new electronic systems, we

13 can automatically and with precision pull those

14 cases out of the queue for fast processing.

15 Our initial test of this concept in

16 New England was highly successful, even though the

17 model only included a handful of cancers, ALS and

18 low birth weight babies.

19 If we do the hard work of identifying

20 the hundreds, perhaps thousands of similarly

21 situated diseases and conditions, which is what

22 you're all here today to help us with, and for

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

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

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

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

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

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

1 current high volume cases automatic and easy.

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

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

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

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

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

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

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

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

20 Steve.

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

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

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

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

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

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

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

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

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

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

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

1 knowledge about their rare disease.

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

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

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

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

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

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

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

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

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

21 So thank you very much.

22 COMMISSIONER ASTRUE: Thank you,

1 Steve.

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

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

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

19 Thank you.

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

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

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

10 COMMISSIONER ASTRUE: Thank you.

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

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

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

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

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

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

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

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

22 And Noni Langford, Chair of the

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

4 Thank you all. Welcome.

5 We will begin with Diane.

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

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

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

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

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

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

13 Rather than discussing the course of
14 individual rare diseases, for the most part, I
15 would like to provide a panoramic, kind of a
16 10,000-foot view of -- of rare diseases.

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

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

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

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

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

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

19 In a survey NORD conducted with
20 Sarah Lawrence College several years ago,
21 42 percent of the respondents said that they were
22 prevented from working because of their disease.

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

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

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

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

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

1 hard. She was 39 years old.

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

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

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

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

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

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

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

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

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

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

13 Thank you.

14 COMMISSIONER ASTRUE: Thank you,
15 Diane.

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

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

3 And we'll move now to Dr. Lipkin.

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

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

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

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

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

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

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

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

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

16 I -- I made a point today of wearing
17 this particular tie. It was designed by a
18 13-year-old -- now a 13-year-old child who I've
19 known since approximately the age of 3. Emily is
20 a happy, smiling 13-year-old child who, in the
21 course of her 13 years of life, has had
22 35 neurosurgical procedures for some of the

1 congenital brain abnormalities.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

22 The pediatrician can provide the

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

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

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

17 We know that children with special
18 healthcare needs make up approximately
19 12.8 percent, or 9.4 million children and youth in
20 the United States.

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

1 child or youth with special healthcare needs
2 living there.

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

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

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

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

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

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

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

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

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

22 We also expect these advances to

1 continue in the coming decades.

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

6 We look forward to your consideration
7 of these factors.

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

11 COMMISSIONER ASTRUE: Thank you.

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

18 Thanks.

19 Sissi.

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

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

5 Thanks for making me feel so welcome
6 here today.

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

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

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

1 promoting public and professional awareness of
2 these disorders.

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

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

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

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

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

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

21 The mucopolysaccharidosis, or MPS,
22 disorders are genetically determined lysosomal

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

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

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

18 My 12-year-old son, Joe, and my
19 11-year-old daughter, Maggie, suffer from
20 Sanfilippo syndrome, or MPS III. It is a result
21 of a genetic recessive mutation that both -- in
22 both mine and my husband's DNA. There's a

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

1 future.

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

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

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

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

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

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

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

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

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

19 Joe's estimate -- estimated budget for
20 the 2006-2007 year is \$69,000, and Maggie's is
21 79,000. That does not reflect the amount of money
22 our private insurance pays out, which is

1 80 percent of the -- of the billing for medical
2 needs.

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

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

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

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

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

1 unable to keep up with the devastating effects of
2 MPS.

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

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

13 They receive about \$300 million in
14 Federal Medicaid funding to provide services.
15 They currently serve 28,000 people. Approximately
16 82 percent of these people live at home with their
17 families, compared to only 60 percent nationally.

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

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

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

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

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

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

1 solutions.

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

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

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

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

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

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

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

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

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

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

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

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

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

1 respond so problems can be resolved quickly.

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

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

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

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

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

1 situation.

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

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

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

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

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

6 COMMISSIONER ASTRUE: Thank you very
7 much.

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

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

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

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

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

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

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

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

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

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

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

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

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

1 Any -- any suggestions or guidance?

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

4 COMMISSIONER ASTRUE: Thank you.

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

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

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

22 DR. LIPKIN: It's an enormous task,

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

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

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

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

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

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

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

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

19 MS. LANGFORD: Sure.

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

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

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

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

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

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

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

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

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

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

3 So although, we're talking about
4 7,000, there is going to be a core that is -- that
5 is absolutely devastating and -- and -- and
6 heartbreaking in many ways and that are probably
7 not going to be quite as difficult to make
8 decisions about.

9 COMMISSIONER ASTRUE: Dave, do you
10 have any?

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

13 UNIDENTIFIABLE VOICE: We can't hear
14 you.

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

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

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

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

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

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

1 that.

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

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

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

1 needs to use that -- those new approaches.

2 ACTING DEPUTY COMMISSIONER RUST:

3 Thank you.

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

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

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

1 reaching out to the family support groups.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

22 And it's important not just for the

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

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

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

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

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

7 Frank, do you have some questions?

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

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

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

22 But it also goes to the -- the

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

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

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

1 as well.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

22 And we're also wrestling with how to

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

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

10 DR. LIPKIN: Right.

11 COMMISSIONER ASTRUE: Any other
12 questions?

13 ACTING DEPUTY COMMISSIONER RUST: No.

14 COMMISSIONER ASTRUE: You got another
15 one?

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

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

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

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

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

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

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

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

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

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

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

4 JUDGE CRISTAUDO: Thank you.

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

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

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

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

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

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

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

1 whatever we can do to -- to assist.

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

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

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

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

16 So thank you very much.

17 MS. DORMAN: Thank you, Commissioner.

18 (Recess.)

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

22 We're going to try to get back on

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

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

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

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

13 Thank you very much for joining us
14 today.

15 MS. LANDRY: Me first.

16 COMMISSIONER ASTRUE: Whichever way
17 you want to go.

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

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

3 MS. LANDRY: Thank you very much.

4 Thank you for this opportunity to testify.

5 As you said, my name is Linda Landry.

6 I'm a senior attorney at the Disability Law
7 Center. I've been representing people before the
8 Social Security Administration for about 27 years.

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

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

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

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

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

1 needs.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

22 I know that most of the DDSs have

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

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

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

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

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

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

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

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

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

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

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

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

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

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

1 available.

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

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

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

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

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

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

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

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

22 But I think there's been some

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

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

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

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

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

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

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

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

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

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

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

5 Thank you.

6 COMMISSIONER ASTRUE: Thank you.

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

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

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

4 So I just wanted to --

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

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

9 Okay, thanks a lot.

10 MS. LANDRY: Thank you.

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

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

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

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

1 Administration.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

8 This would allow people with ALS to
9 get into the Medicare system sooner than the
10 24 months that is typically required, and that's
11 because typically, they don't survive 24 months.
12 Now, typically by the time they qualify under
13 the -- the old law, the Medicare card came too
14 late for them.

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

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

22 Part of the problem, you know, we --

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

1 all it says.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

22 I do sometimes have some -- some

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

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

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

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

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

1 they're actually entitled to.

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

4 COMMISSIONER ASTRUE: Thank you.

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

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

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

18 I would say I'm in the recovery from
19 the practice of law. Instead, I do the advocacy
20 work. And I've been speaking around the country,
21 basically every other weekend, for the last
22 13 years for many, many groups of people who need

1 disability benefits.

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

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

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

1 couldn't.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

1 lower appeals.

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

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

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

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

1 Social Security Administration in making decisions
2 well and quickly.

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

22 These consumer advocates could also

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

18 COMMISSIONER ASTRUE: Thank you very
19 much.

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

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

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

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

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

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

22 MS. LANDRY: I -- I think that it

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

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

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

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

1 their -- their cooperation.

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

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

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

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

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

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

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

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

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

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

7 In terms of trying to get the specific
8 information --

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

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

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

18 MS. LANDRY: They can charge.

19 COMMISSIONER ASTRUE: They can charge
20 them?

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

1 COMMISSIONER ASTRUE: And what's --

2 and what typically is the charge?

3 MR. LEACH: It's --

4 MS. BERNSTEIN: Varies.

5 MR. LEACH: -- it varies a lot.

6 Hospitals tend to be very high. They'll want to
7 do a per page charge, and hospital records tend to
8 be voluminous. I just paid -- or not me,
9 personally, but our program just recently paid
10 something upwards of \$300 for medical records for
11 a child.

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

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

22 COMMISSIONER ASTRUE: Well, why don't

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

4 Yeah, we have a historic church right
5 next door.

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

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

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

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

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

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

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

21 COMMISSIONER ASTRUE: Okay.

22 MS. LANDRY: Absolutely.

1 COMMISSIONER ASTRUE: David, you got
2 anything?

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

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

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

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

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

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

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

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

1 these conditions overlap.

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

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

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

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

22 But I do want just to mention, too,

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

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

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

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

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

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

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

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

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

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

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

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

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

10 DR. GROFT: Thank you.

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

14 COMMISSIONER ASTRUE: Little closer to
15 the mike.

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

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

22 Ms. Landry, I think you may have said

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

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

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

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

1 information.

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

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

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

1 on a consistent basis without too many absences.

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

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

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

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

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

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

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

1 recurring cases.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

1 and how many days does it take afterwards.

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

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

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

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

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

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

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

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

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

7 Dave, any more?

8 Frank?

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

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

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

1 (Laughter.)

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

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

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

10 Thank you.

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

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

2 (2:51 p.m.)

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

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

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

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

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

22 Thank you, gentlemen.

1 I guess we'll start with Walter.

2 DR. KOROSHETZ: Thank you very much.

3 It's a pleasure to be here, Commissioner Astrue,
4 and members of the committee. So I'm representing
5 the National Institute of Neurologic Disorders and
6 Stroke.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

1 brain disease.

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

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

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

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

1 determinations, it raises concern.

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

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

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

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

1 disease.

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

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

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

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

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

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

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

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

22 Fronto-temporal dementia affects the

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

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

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

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

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

1 that -- how disabled they were.

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

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

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

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

1 limited.

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

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

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

1 specific disease.

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

8 All who attended realized the
9 difficulty inherent in making 100 percent accurate
10 determinations on the first attempt in all
11 2.6 million applicants a year. But they're
12 attracted by the possibility that at least being
13 able to make sure that a significant number of --
14 of severely disabled patients could be identified
15 in -- with some type of formulaic approach.

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

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

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

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

7 So thanks very much for your
8 attention.

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

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

17 MR. GIBSON: Great. Thanks.

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

22 I appreciate the opportunity to speak

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

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

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

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

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

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

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

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

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

1 breathe. This is a horrific disease.

2 The average lifespan for a person with
3 ALS is two to five years from the time of
4 diagnosis. However, the disease progresses
5 differently in different people, and about
6 50 percent die within 18 months of diagnosis.
7 This -- there is no known cause, cure or means of
8 prevention for this disease.

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

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

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

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

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

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

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

1 for sudden loss of income.

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

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

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

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

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

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

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

1 Administration.

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

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

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

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

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

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

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

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

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

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

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

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

11 Four, additional problems are
12 encountered by people with ALS not related to the
13 disability, but to work requirements like the
14 20/40 rule and the five-month waiting period.

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

18 Ultimately, most of the -- the --
19 these cases are resolved favorably, and since
20 2003, we, in our chapters, have encountered fewer
21 instances of difficulties.

22 It should be noted that there are

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

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

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

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

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

1 for presumptive disability payments under SSI.

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

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

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

18 We have a few suggestions.

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

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

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

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

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

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

1 assistance in serving these constituencies.

2 Indeed, we welcome opportunities to educate local
3 offices about ALS and to provide information to
4 them that would make their jobs easier and reduce
5 problems to expedite claims.

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

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

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

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

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

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

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

22 We believe SSA should consider

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

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

14 COMMISSIONER ASTRUE: Thank you.

15 We'll move to Ron.

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

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

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

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

22 So we are wonderfully excited in our

1 community about the prospects for treatment.

2 We're getting five clinical trials,
3 maybe even six over the course of the next
4 12 months. And we are absolutely convinced that
5 insights gained and benefits gained from those
6 clinical trials will be beneficial for other
7 disease groups that you're dealing with today and
8 in your work.

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

14 And so let me -- let me say in that
15 regard that our son, Keith, has a rare,
16 life-shortening genetic disorder called
17 Friedreich's Ataxia.

18 Though Keith was born a healthy
19 infant, as was the case with many of the panelists
20 today, in December of 1985, by the time he was
21 8 years old and in the 3rd grade, he began to
22 stumble and fall for no apparent reason.

1 He also began to have increasing
2 difficulty writing legibly. His hands were
3 cramping and so were his legs. These symptoms
4 worsened to the point that when Keith was
5 11 years old, we had him tested, and he was
6 genetically confirmed to have Friedreich's Ataxia.

7 Like most parents who receive this
8 diagnosis, we had never heard of
9 Friedreich's Ataxia. I wish I could still say
10 that. We learned a lot about the disease, though,
11 on the day of diagnosis.

12 We learned that Friedreich's Ataxia is
13 a neuromuscular disorder inherited by way of a
14 recessive genetic mutation carried by about 1 in
15 90 people. We learned that when two such carriers
16 of -- have children, each offspring has a
17 one-in-four chance of inheriting the mutation from
18 both parents and being afflicted with the
19 disorder.

20 We learned that Friedreich's Ataxia is
21 relentlessly progressive and would soon deprive
22 Keith of strength and coordination in all four

1 limbs, placing him in a wheelchair by the time he
2 was a teenager.

3 We learned that Keith's vision,
4 hearing and speech would all be diminished; that
5 he would develop severe scoliosis, or curvature of
6 the spine, requiring surgical implantation of
7 metal rods to straighten it, a horrific and brutal
8 surgery; that he would be at greater risk of
9 developing diabetes; and that he would most likely
10 have a severe heart condition.

11 Finally, we learned that there's no
12 treatment or cure for Friedreich's Ataxia, that
13 average life expectancy for Friedreich's Ataxia
14 patients is early adulthood.

15 Within -- within just a few weeks of
16 Keith's diagnosis, after a number of medical
17 examinations, we knew he already had a severe
18 heart condition and the early stages of scoliosis.
19 His weakness and incoordination progressed
20 rapidly, and he began using the wheelchair
21 full-time when he -- when he was 16.

22 His scoliosis also worsened, and we --

1 and he required the spinal fusion surgery to
2 correct his spine that same year.

3 His vision, hearing and speech
4 steadily declined and continue to do so. He
5 developed Type 1.5 diabetes at age 20.

6 So by the time Keith turned 18 and was
7 eligible, or we hoped eligible to apply for SSI as
8 an adult, he had been in a wheelchair for more
9 than two years, had endured spinal fusion surgery,
10 could not speak clearly, and his vision and
11 hearing were both impaired.

12 He had an individual education plan
13 for the final seven years of his education. Keith
14 had worked hard and done well in school, despite
15 his disabilities, and graduated from high school
16 with his class after posting a 4.0 grade point
17 average during his senior year.

18 Although he went to our local
19 community college for several semesters and did
20 well in his classes, the relentless progression of
21 this disorder left him extremely fatigued and made
22 it increasingly difficult to read and keep up with

1 his assignments.

2 Transportation was another
3 complicating factor in that Keith had never been
4 physically able to drive, so had to rely on others
5 to get him around.

6 With his physical abilities and the
7 steep decline and the demands of his education on
8 the rise, Keith reluctantly decided he could not
9 continue his schooling.

10 He was also disappointed to find that
11 he was unable to find a job.

12 Again, because Friedreich's Ataxia is
13 relentlessly progressive, Keith's physical
14 capabilities have continued to decline steadily to
15 the point of needing around-the-clock assistance
16 for even the most basic needs.

17 The diabetes associated with
18 Friedreich's Ataxia carries its own additional
19 requirements in this regard. Keith is unable to
20 perform the blood glucose testing or administer
21 his own injections, for example, both of which are
22 required several times each day.

1 With all of these increasing
2 challenges, you can understand how welcome to our
3 family was the very quick SSA decision on Keith's
4 disability claim. You can imagine how encouraged
5 Keith and we were when he received, for example,
6 his first SSI check even before he re- -- received
7 his approval letter.

8 With no other income or resources,
9 Keith had no other way to support himself. The
10 SSI monthly payment helps Keith pay for rent,
11 food, utilities and personal care items. It is
12 critical for Keith to have the financial ability
13 to take care of his basic needs.

14 Fortunately for Keith and us,
15 Friedreich's Ataxia is one of the rare disorders
16 listed in Section 11.17 of the disability
17 evaluation under the Social Security Blue Book of
18 June 2006. Because of Keith's positive
19 experience, he and we now realize that not only is
20 the SSI program extremely important in and of
21 itself, SSI approval also provides a gateway to
22 applying for additional social services, for

1 example, Medicaid.

2 Because Keith was approved for SSI, he
3 was able to apply for Medicaid through the
4 Commonwealth of Virginia. He met the Federal
5 income and asset eligibility standards, and
6 Medicaid helps pay for his doctor and hospital
7 care, medication and durable medical equipment,
8 such as his wheelchair.

9 Home base care. Keith has continued
10 to decline dramatic -- dramatically in his ability
11 to take care of himself. Personal grooming,
12 dressing, meal preparation and eating, just to
13 name a few tasks, he now finds very difficult or
14 impossible.

15 With SSI and Medicaid approval in
16 place, Keith was evaluated and found eligible for
17 personal care services. He receives caregiver
18 services five hours per day, Monday through
19 Friday. The caregiver helps him bathe, dress,
20 prepare his meals, washes his clothes and vacuums
21 his floor -- vacuums his room. Not only is this
22 of benefit to Keith, it allows us, my wife and I,

1 daily -- a daily block of time for our own
2 employment and time to care for our other family's
3 routine needs.

4 Respite care. When Keith's caregiver
5 is not with him, either his mom or I have to be
6 available to take care of his needs. This reality
7 has become part of our daily lives. We love our
8 son and want to help him in any way we can, but we
9 realize at the same time, like so many others, we
10 will not be able to help take care of Keith unless
11 we take care of ourselves and our other
12 obligations.

13 Keith and we were grateful, therefore,
14 that Keith was granted approval of preauthorized
15 service for respite care in the amount of
16 720 hours per year. Respite care helps to refresh
17 us, to sustain ourselves and our family. It helps
18 us take care of ourselves and our family so we can
19 take better care of Keith.

20 I doubt, Mr. Commissioner, that anyone
21 will come before you and say that it is easy
22 living with a rare disease or helping care for

1 someone with a rare disease.

2 I would like to assure you, though,
3 that the SSI program helps many people, including
4 our son, both directly and indirectly, in tangible
5 and intangible ways.

6 It helps directly by providing him the
7 wherewithal to provide for himself as much as
8 possible. It helps indirectly by -- by providing
9 a valid foundation that opens the door to
10 important additional social services.

11 It helps in the tangible physical ways
12 I've outlined. It helps in important intangible
13 ways, too. It reinforces the recipients self --
14 sense -- sense of self-sufficiency, self-esteem
15 and dignity. It works in all these ways to help
16 provide the recipient with a higher quality of
17 life than otherwise could be achieved.

18 I applaud the Social Security
19 Administration in reviewing the need for
20 compassionate allowances in an attempt to
21 streamline this system for other patients with
22 rare diseases, and I look forward to assisting in

1 any way I can.

2 In terms of your efforts to develop
3 the most effective methods for identifying rare
4 medical conditions appropriate for compassionate
5 use allowances, I know that you're already engaged
6 in most, if not all, of the following
7 undertakings, so I will simply emphasize them for
8 the vantage point of our own very positive
9 experiences.

10 One, I would suggest, for example,
11 continuing to work closely with your colleagues at
12 the National Institutes of Health, obviously, as
13 the membership of this panel indicates you are
14 doing.

15 More specifically, I would suggest
16 that you continue to work closely with the
17 NIH Office of Rare Diseases, represented so ably
18 by Steve Groft, as well as with each of the NIH
19 institutes responsible for each of the rare
20 diseases under review.

21 Again, the list of witnesses for this
22 hearing makes clear that you are on that case, so

1 to speak.

2 I had the good fortune to serve for
3 four years on the National Advisory Committee of
4 one of those NIH institutes, the National
5 Institute for Neurological Disorders and Stroke,
6 represented here on this panel by its deputy
7 director, Dr. Koroshetz.

8 And I would like to join the millions
9 of others in applauding the NIH as the world's
10 leading funding agency for medical research. You
11 are no doubt finding at the NIH highly qualified
12 scientists who can help identify the trigger
13 point, so to speak, within rare diseases that can
14 serve to validate speedy SSI approval.

15 If there is anything you believe a
16 patient advocate like myself might be able to do
17 to be of assistance in this regard, in a liaison
18 capacity, for example, I and many others would be
19 happy to work with SSA in any way that might prove
20 helpful.

21 As president of the Friedreich's
22 Ataxia Research Alliance, I would also like to

1 suggest that you continue to work closely as you
2 are with nonprofit advocacy organizations that
3 support research and services for patients with
4 rare diseases.

5 Such organizations have formed around
6 many rare diseases and would be happy -- more than
7 happy to help wherever they can.

8 The National Organization of Rare --
9 Rare Disorders, NORD, which was represented ably
10 this morning on a panel, is represented on -- is
11 also clear -- clearly ready to help.

12 To illustrate the same point using our
13 own organization, FARA, we frequently answer
14 questions and provide advice to patients who are
15 preparing to file an application for SSI. We help
16 them, for example, in -- in identifying the
17 employment, education and medical documentation
18 required for filing with the other Social --
19 specific Social Security regulations pertaining to
20 our disease and in using the online screening
21 tool, called BEST.

22 Rare disease patient advocacy groups,

1 as you pointed out in front of an earlier panel,
2 tend to know a lot about their disorders in part,
3 as you pointed out, because they have to educate
4 themselves as well as many of their family
5 physicians who didn't learn much about these rare
6 diseases in medical school and have had precious
7 little contact with them since.

8 Such advocates and -- and the
9 scientists with whom they are closely aligned
10 could offer significant help in identifying the
11 trigger points for compassionate allowances and
12 these rare diseases, and they can be extremely
13 helpful in educating the rare disease patient
14 community in terms of the process involved in
15 applying for SSA benefits.

16 For example, going back to the
17 excellent exchanges you've had this morning with
18 previous panels, the medical advisors working with
19 such advocacy groups could provide invaluable
20 insights and potential function -- on potential
21 functional criteria. In the case of a number of
22 rare diseases, for example, clinical networks that

1 they've helped develop -- that the advocates have
2 helped develop, are working hard to develop
3 clinical measures and scales essential to
4 determining progression of disease, as well as
5 therapeutic effect of any therapy to be tested in
6 a clinical trial.

7 These measures and the scales behind
8 them are fine tuned to the specific disorders in
9 question and would include validated
10 questionnaires for quality of living and
11 activities of daily living.

12 Like the -- the MELD test that -- or
13 scores that you mentioned this morning,
14 Commissioner Astrue, for -- for liver, there are
15 such scores being developed for not just
16 individual rare diseases, but in many cases, for
17 collections of rare diseases that answer to the
18 same underlying mechanisms of damage and,
19 therefore, might respond to the same underlying
20 mechanisms of action for therapeutics.

21 So I think that's a really promising
22 avenue of approaching the advocate, the NIH,

1 the -- the rare disease groups and their
2 scientists and advisors, because some of those
3 measures and scales and -- and scores are being
4 developed.

5 In many cases, the request -- first,
6 currently, the SSI application process requests
7 very generally medical reports, and in many cases,
8 that request for general medical reports is
9 adequate. It wasn't ours, because we had -- we
10 know our gene, we know -- we have a genetic
11 confirmation test, we know a lot about our -- our
12 symptoms, and it's relentlessly progressive. And,
13 in most cases, the applicants for SSI are already
14 in their wheelchairs. You know, they're already
15 clearly able to qualify under the single
16 functional capacity which is, in our case, quote,
17 significant and persistent disorganization of
18 motor function in two extremities, resulting in
19 sustained disturbance of gross and dexterous
20 movement or gait and station, end quote.

21 Our son was already in his wheelchair,
22 as most of your other applicants are; he was

1 already unable to communicate clearly, clearly met
2 those -- that functional capacity criterion.

3 However, in cases of later onset of
4 our disease -- there are cases of later onset, so
5 that the adult doesn't develop the symptoms until
6 mid 20s, sometimes even later, that would not be
7 so easy a call.

8 And so in that case, the process that
9 you've already outlined of consulting so broadly,
10 as you already are doing, can further instruct
11 the -- the process and -- specifically by asking
12 more -- for more and better information at the
13 outset, more and better information in the early
14 stages, rather than waiting for the hearing stage
15 two years later.

16 At the application process, if -- if
17 the list and the functional criteria -- criteria
18 are instructed by the kinds of things you will
19 learn in these consultations, we believe that --
20 and -- and maybe even the measures and scales
21 and -- and test scores that -- that -- that you're
22 looking for are there, available to the very

1 helpful local representative of SSA -- in our
2 case, extremely helpful, sat down with us, helped
3 us fill out the application, gave us some guidance
4 on where to go to get these medical reports.

5 But if -- if that local representative
6 was -- had the additional information in front of
7 her or him to be able to tell the parent and the
8 patient, you know, this is the kind of medical
9 report you need and -- and if you've -- if you've
10 gone to a clinic that has a specific examination
11 for your rare disease, can you get a test score?

12 And -- and, you know -- those -- those
13 test scores are increasingly available in a -- in
14 a -- in a number of rare diseases like ours, and I
15 think that will continue to be the case as you do
16 your work.

17 So in -- in -- in sum, I -- I believe
18 the kinds of collaborations you are already
19 discussing with -- with your panels and -- and
20 represented by your colleagues on your own panel
21 and -- that productive collaboration is -- is
22 already afoot and -- and that you will benefit

1 tremendously and that -- that collaboration will
2 be extremely productive and result in more and
3 better information early in the process.

4 In final conclusion, thank you very
5 much for the benefits of the SSI program. They
6 provide to our -- our son and many others in our
7 families a -- a great deal of benefit, both
8 tangible and intangible.

9 Thank you also for your efforts to
10 make SSI available quickly to the other families
11 living with such devastating rare diseases through
12 the proposed compassionate allowances initiative.
13 And it's my pleasure and honor to be here.

14 COMMISSIONER ASTRUE: Thank you.
15 Thank you very much. Thank you to the entire
16 panel.

17 I've got a -- a few questions here.

18 Both these diseases are ones that I
19 worked on professionally in the past, and so, you
20 know, I don't have to be persuaded on, you know,
21 what these disease are like.

22 So I've just got a couple technical

1 questions.

2 I wanted to ask you about ALS in terms
3 of the difficulty of diagnosis. And if you could
4 maybe walk me through a little bit what your
5 experience has been in terms of what it tends to
6 get confused with and then how that gets sorted
7 out, because if we know that, that might make it a
8 little bit easier for us to be clearer in our
9 standards when we're trying to sort that out to
10 make sure -- because the question for ALS is
11 really just do we have a legitimate diagnosis?

12 As you know, you're in -- ALS is in
13 the QDD program now, and last week I think we were
14 up to 28 states, we should be 50 states before
15 very long, and they should be routinely allowed
16 within sort of 10 to 14 days very shortly. So, I
17 mean, there is some real progress going there.

18 But it seems to me in the case of ALS,
19 the big progress is just making sure that we're
20 asking the right questions to make sure that we've
21 got the right diagnosis.

22 So if you could walk me through that a

1 little bit --

2 MR. GIBSON: Sure.

3 COMMISSIONER ASTRUE: -- that would be
4 helpful.

5 MR. GIBSON: And we'd be happy to
6 provide the Agency with a more medical/scientific
7 background. I'm a government relations
8 professional, I can tell you what I've experienced
9 in my 10 years of the office here in Washington.

10 As I said in my remarks, there is not
11 one test, so basically a person with ALS goes
12 through a series of -- of tests and various
13 doctors' appointments.

14 We've had stories of patients coming
15 in, first being diagnosed as carpal tunnel
16 syndrome, where they actually have not had
17 movements in their hands. We've had situations
18 where it's confused with Lyme's disease, where
19 there's some connections.

20 COMMISSIONER ASTRUE: Probably
21 Guillain-Barré and fibromyalgia and things in that
22 class, too?

1 MR. GIBSON: Right, right. Correct.

2 And sometimes there is some confusion
3 with MS. Generally what happens is ALS is -- is a
4 death sentence. So any sort of medical
5 professional does not like to give that diagnosis
6 until everything else is ruled out. But, you
7 know, a lot of times -- we have stories of an
8 actor who was on a soap opera who was fired from
9 his soap opera because they thought he was
10 actually using drugs because he couldn't negotiate
11 certain words, and that went through a whole legal
12 ramification with Proctor and Gamble where they
13 finally diagnosed him with ALS, and that took two
14 and a half years.

15 But we'd be happy to give a list for
16 all the agencies to look at for various examples.

17 COMMISSIONER ASTRUE: Yeah. Well,
18 maybe you -- you can update us.

19 DR. KOROSHETZ: Yeah, it's a -- you
20 know, there's -- like in many different
21 neurological diseases, there are a variety of
22 presentations, and the classic ALS presentation of

1 weakness, atrophy of muscles, vesiculations in the
2 muscles with confirmation with
3 electrophysiological test, the EMG, you can make a
4 fairly significant, you know, diagnosis.

5 You'd like to exclude, of course,
6 anything that's treatable because of ALS not
7 having a treatment. There are some disease --
8 disorders that can mimic ALS, but those -- I think
9 those kind of sort themselves out.

10 The ones that are tricky are the
11 ones -- as -- as Steve mentioned, for instance, do
12 they present bulbar-wise. So it's primarily
13 speech, and there is no weakness, so somebody is
14 saying, well, how could you have ALS if you're not
15 weak, and there's a bulbar form that you have to
16 know about.

17 Or there's another form, which is
18 progressive lateral sclerosis, where the primary
19 problem is actually in the motor cortex, and the
20 patient develops incoordination and stiffness but
21 not that much weakness. So those are the kind of
22 variations that sometimes will cause someone not

1 to be diagnosed.

2 There's another form where it affects
3 the respiratory muscles, and they present with
4 shortness of breath. Sometimes they end up in a
5 medical unit because they're having trouble
6 breathing, and that -- that can be a real tricky
7 one to sort out.

8 COMMISSIONER ASTRUE: Is -- is there a
9 time -- now the -- the time -- usual time from
10 diagnosis to death is just a few years, usually,
11 right? Is there -- from the time where there --
12 there -- there's an attempt to diagnose to when
13 you really have a -- a confirmed diagnosis, does
14 that tend to be three months? six months?
15 twelve months?

16 DR. KOROSHETZ: You know, it's on an
17 average -- you know, my rule is try never to make
18 that kind of diagnosis the first time, always give
19 a chance to see what happens over time. I'd say
20 an average is somewhere around 3 months if you're
21 in a -- you're in a system with good neurology.

22 MR. GIBSON: But I think that varies

1 on where you live in the U.S. If you're in a
2 rural area where you might not even have a
3 neurologist in the vicinity, you have to then
4 travel on.

5 We have, you know, many folks in the
6 Dakotas and Montana have to go to Mayo, so I think
7 that can take months to get another appointment,
8 which then leads to a year and a half or so.

9 COMMISSIONER ASTRUE: Okay. I had
10 another -- another question for Ron, and again,
11 I -- I realize, consistent with Steve's response,
12 I may be asking too much medical, and so if -- if
13 there's anything you want to submit for the record
14 afterwards from the -- from the Alliance, I'd
15 be -- be happy to take it.

16 I have some general recollection that
17 Friedreich's Ataxia is sort of the most common in
18 sort of a loose family of somewhat similar
19 diseases, most which of are even rarer than
20 Friedreich's Ataxia.

21 Could you may -- tell me maybe a
22 little bit more about the -- the other ataxias and

1 whether you have any observations about the
2 problems that they have in terms of diagnosis and
3 system -- and -- and -- and going through our
4 system and other systems as well?

5 MR. BARTEK: I will give it my best
6 shot, Commissioner Astrue, and -- and -- and
7 provide some additional materials later.

8 But, yes, you're referring to a
9 general family of ataxias, and there are on the
10 order of two dozen or a few more now, maybe up to
11 30 different identified ataxias. The vast
12 majority of them are dominant trait ataxias.

13 There are only probably two or three
14 recessive trait ataxias like Friedreich's Ataxia
15 and Ataxia telangiectasia. And the -- and, yes,
16 you're right, Friedreich's Ataxia's is by far the
17 most common of those ataxias.

18 Roughly speaking, Friedreich's Ataxia
19 patients constitute about half of the ataxia in
20 the population, all the others combined being
21 about an equal number.

22 They -- about -- I'll venture a guess

1 that there are roughly a third to a half of those
2 ataxias now that have genetic confirmation tests
3 available, and leaving the other half without any
4 specific genetic marker. And the majority of
5 those with specific genetic confirmation are, like
6 Friedreich's Ataxia, triplet repeat expansion
7 diseases, polyglutamine ataxias, the difference
8 being that those triplet repeat expansions are on
9 coding regions, exons of their genes, so they --
10 they -- they do a gain of function problem so that
11 the protein produced by those genes are
12 problematic.

13 In our case, our expansion, believe it
14 or not, is an intron, and so every time we get a
15 pass, an effective pass, we get effective protein,
16 we get a perfectly formed frataxin protein. So
17 that introduces differences in therapeutics,
18 obviously.

19 But -- so that -- that's a -- an
20 attempt at summarizing an answer to your
21 question.

22 COMMISSIONER ASTRUE: What --

1 what would probably be helpful to us, I -- I --
2 you know, I know that our folks are -- are -- are
3 on top of Friedreich's Ataxia. What I don't know
4 is, for the whole rest of the family, exactly
5 where they are and if they're up -- you know, up
6 on definitive diagnosis and that kind of -- if --
7 if you could submit --

8 MR. BARTEK: Yes.

9 COMMISSIONER ASTRUE: -- that for the
10 record. Again, it may be that I'm not giving them
11 enough credit, but it certainly wouldn't hurt to
12 have the most recent analysis. If you have that,
13 that would be great.

14 MR. BARTEK: I -- I -- I will do that,
15 and I'll say at this point that there's a great
16 deal in common in terms of symptoms. Ataxia is a
17 Greek word, as you well know, meaning
18 incoordination, and there is plenty of
19 incoordination amongst all two dozen disorders.

20 A key difference, though, is -- is age
21 of onset. For the most part, the dominant
22 ataxias, the spinal cerebellar ataxias, are much

1 later onset. Our onset is usually between the
2 ages of 5 and 15. There are -- there are these
3 cases of later onset Friedreich's Ataxia where the
4 onset can be in the 20s, 30s, maybe even later.
5 So --

6 COMMISSIONER ASTRUE: Is -- Ron, is
7 there any -- can you tell from genetic testing
8 between the -- the early onset and later onset or
9 do they appear the same in the gene?

10 MR. BARTEK: There are some markers
11 that are generally true. For the most part, you
12 know, again, it's -- these are triplet repeat
13 expansion disorders, and the size of the expansion
14 in the shorter allele is going to be usually
15 indicative of age of onset, speed of progression,
16 severity of final outcome and nonmandatory
17 symptoms. Nonmandatory symptoms include the heart
18 condition, diabetes and -- and so forth.

19 So the lower the expansion, you know,
20 the shorter the expansion in Friedreich's Ataxia,
21 because it is in the intron, the -- the assumption
22 is the more frataxin protein you get, and so

1 the -- the later the onset.

2 So that could be a helpful, you know,
3 sort of designator or marker, if you will, for the
4 later onset patients that will eventually -- it is
5 relentlessly progressive, so at some point, they
6 will be eligible in almost every case. But I know
7 you'll have to determine, you know, when -- when
8 the onset is sufficient to -- to trigger the
9 benefits.

10 COMMISSIONER ASTRUE: And that's a
11 good point. And that actually triggered me to --
12 to just sort of say something, so I'm not going to
13 pretend that everything's a question.

14 One thing that is important for anyone
15 in the audience that is trying to help us help you
16 to remember is what we're -- we've got this push
17 to look at imaging data and biomarkers and -- and
18 try to draw distinctions in that way.

19 The statute also is very specific that
20 we're actually not only allowed, but required
21 when -- when it's appropriate, to use age in
22 making these -- these types of judgments, too. So

1 in many cases, many diseases, that's not very
2 helpful, but there are certain degenerative
3 diseases where the natural history is arguably so
4 predictable that we might be able to use age as a
5 factor, too.

6 So if you're -- you're thinking a --
7 about ways to help us draw the line in an
8 appropriate place, don't over -- don't overlook
9 age, because there might be -- probably relatively
10 few diseases and conditions, but there might be a
11 few where the natural history is sufficiently
12 consistent that we might be able to use that as a
13 fuller or partial factor in a decision.

14 All right. I've been hogging air
15 time.

16 Anybody to my left have a question?

17 DR. GROFT: Yeah. I -- I've got a
18 question for Ron or Steve. You both have a
19 considerable amount of information available about
20 the individual diseases that -- that you
21 represent. And I think we heard this morning in
22 earlier panels about needing more information,

1 maybe different information.

2 I guess the question is: How
3 difficult, from what you've heard today, would it
4 be -- and, Ron, you've done through the process
5 personally -- how difficult would it be to recast
6 the information that you currently have available
7 and to make it information that would be useful to
8 the Social Security Administration?

9 MR. BARTEK: I don't think it would be
10 difficult at all, Steve, and we'd be eager to do
11 that. We -- we are fortunate in having a clinical
12 research network already established, a network
13 that's already seen several hundred of our
14 patients, have developed over a three-year period
15 natural history progression against these ataxia
16 scales, if you will. That includes such things as
17 a 25-foot walk, with or without assistance, you
18 know, a time to walk, nine-hole pegboard test,
19 vision testing and speech testing and -- and so
20 forth.

21 So these scales are readily available.
22 We could provide information that -- that's a

1 national history for these patients -- that --
2 that set of patients that I think would be
3 extremely helpful.

4 MR. GIBSON: Yeah, it would not be
5 difficult at all. Back in 2001 -- actually, 2000,
6 when Congress passed the Medicare waiver for
7 people with ALS, we started to develop a
8 partnership with -- not only with the SSA here
9 nationally, but throughout the country, and we'd
10 be happy to continue that partnership on giving
11 the information.

12 COMMISSIONER ASTRUE: I should -- I
13 should note, too, one -- you know, one of the
14 reasons why we're drawing two categories, the
15 compassionate allowance and the QDD, for a lot of
16 the people that are very compelling, we can't make
17 a complete judgment, so we have to go through our
18 full, you know, process to evaluate function.

19 One of the advantages of having the
20 compassionate allowance category -- and, again,
21 we -- we haven't made final decisions on our
22 business processes and forms, and -- and that may

1 be a work in progress for some time, but it at
2 least opens up the possibility that we can do a
3 short form since -- since really, for
4 compassionate allowances, if we're operating on
5 a -- on a conclusive presumption, all we really
6 need to do is confirm that someone is eligible for
7 Social Security and whether they're Title II or
8 Title XVI and -- and confirm the diagnosis.

9 We don't necessarily need all the
10 other information that's going to be relevant for
11 the decision.

12 Likewise, in most, but not all,
13 states, we have rules that require both an
14 examiner and a medical professional to review a
15 case. We have, in some states, what we call
16 single decisionmaker. If we're operating on
17 basically a conclusive presumption, we also ought
18 to be able to cut out one level of review, and --
19 and I -- I would think go to single
20 decisionmakers, too.

21 And that's -- that is something that
22 we've heard from a lot of the states which are

1 really genuinely enthusiastic about this whole
2 endeavor.

3 So both of you, I think, have touched
4 on it briefly, and I just want to say, we are
5 trying to be responsive to that. I don't
6 know that we -- with all the systems changes that
7 we have to make and all the pressure we're under,
8 I'm not sure that we'll have that the way we'd
9 like to have it eventually when we try to do the
10 first rollout next year. But it's certainly our
11 aspiration to make this as -- make application,
12 particularly for people on compassionate
13 allowances, as easy as possible.

14 So -- David, do you have anything,
15 or --

16 ACTING DEPUTY COMMISSIONER RUST: Just
17 two quick ones.

18 Mr. Bartek, you had mentioned -- and
19 this is something you'll probably want to submit
20 to us as -- as opposed to trying to answer
21 today -- but you mentioned that your -- your
22 organization or organizations are beginning to

1 develop numerical metrics.

2 We'd be very interested in seeing
3 where that is and what you've developed, so if you
4 could submit some information on that, I think
5 that might be very helpful to us in crafting this
6 initiative.

7 MR. BARTEK: Thank you. I'll -- I'll
8 look forward to doing that, and it won't take long
9 because we -- we've been developing these metrics
10 for about nine years and -- and we've worked very
11 closely with the NIH in doing so, and we even have
12 them in electronic form.

13 So it's -- and we -- we have what we
14 call a FARS score, which is comparable, in some
15 ways, to the MELD score that the Commissioner was
16 talking about. And -- and we -- there will
17 probably be a numeric metric there that says if
18 you -- if your FARS score is greater than 22, you
19 know, you're eligible.

20 You know, I -- I think --

21 COMMISSIONER ASTRUE: That -- that
22 potentially could be important. I mean, obviously

1 not just for your disease, but --

2 MR. BARTEK: Exactly.

3 COMMISSIONER ASTRUE: -- for a whole
4 wide range of people. So we'd be very interested
5 in seeing that.

6 MR. BARTEK: Yeah.

7 And, in fact, let me say that in that
8 regard, that large portions of that scale that
9 produces that score were -- were borrowed from the
10 MS scales, and portions are shared in common with
11 the Huntington's disease community and -- so I
12 think there -- this will be a pocket of disorders.
13 And including all the other ataxias, you know,
14 that at some point, you know, these kinds of
15 scales will be productive of -- of a score that
16 will be very helpful.

17 COMMISSIONER ASTRUE: Good.

18 Yeah.

19 DR. KOROSHETZ: I'd just say -- that,
20 you know, in trying to develop treatments for
21 diseases, the first step is to develop a scale
22 whereby you can follow patients. So these scales

1 are all kind of built out of consortia that get
2 together to try and develop a method of knowing if
3 a disease is affected by a treatment. And some of
4 them, like in Parkinson's, there's probably 10,000
5 people who've been scaled up, or Huntington's.

6 The rare diseases are -- may not have
7 the same numbers, but any disease where you're
8 trying to get a treatment, they're going to first
9 have to get a scale, and the issue is can you then
10 correlate a score on the scale with inability to
11 work.

12 COMMISSIONER ASTRUE: Right.

13 DR. KOROSHETZ: And, you know, if
14 you've done 10,000 people, the -- the data is out
15 there somewhere. If -- you know, if we can
16 capture it, it might be a way of going quickly to
17 a disability determination.

18 COMMISSIONER ASTRUE: And -- and we
19 have -- we've stopped a couple of things in
20 process. We've got a little room in our R&D
21 budget. We've never actually run studies like
22 that before to try to correlate those types of

1 data with functionality, but we're, as you know --
2 you know this because we've been hounding you, but
3 most people in the room don't know that we're very
4 keen to try to start doing this, and we realize to
5 do it right, we may have to do some investment in
6 our -- from our R&D portfolio, which is a new
7 practice for us, but we're -- we're committed to
8 doing that.

9 So --

10 ACTING DEPUTY COMMISSIONER RUST: And
11 -- and we also have the ability to pull cases and
12 look at what the outcome of the -- of the --
13 the -- the determination process was vis-a-vis
14 those scores and give us some idea of how those
15 scores correlate to disability, to a determination
16 of disability, so --

17 I was also going to thank

18 Dr. Koroshetz --

19 COMMISSIONER ASTRUE: You may -- you
20 may need to get closer to the mike.

21 ACTING DEPUTY COMMISSIONER RUST:

22 Excuse me. I also want to thank Dr. Koroshetz for

1 his -- his help in -- in getting this initiative
2 off the ground and just ask you, you know, in our
3 discussions in August, when you had a number of
4 your colleagues, there was some interesting ideas
5 that were surfaced again about the idea of -- like
6 with schizophrenia plus a cognitive test, the --
7 the diagnosis plus a cognitive test might -- might
8 lead to a determination.

9 I was just wondering, are those
10 discussions still going on? Have you been engaged
11 in any of those around the office?

12 DR. KOROSHETZ: No, I think, as you
13 said, the -- the issue of -- I think is what the
14 common theme that's -- a lot of people come up
15 with is, you know, the diagnosis is not enough,
16 and in some cases, it is, as you mentioned,
17 Commissioner Astrue, the diagnosis determines
18 disability.

19 In a lot of cases, it's a -- it's a
20 match between the diagnosis and the severity of
21 illness, and the question is then how do you
22 measure severity of illness?

1 The Social Security forms that the
2 doctors fill out is kind of a generic way of doing
3 it, and to -- to the issue that I brought up is
4 that if you -- you know, if you want to go into
5 the rare diseases, you get so far -- so far with
6 the generic determination, but you may need to --
7 you know, to have a -- a -- a -- a specialized
8 view towards some of these rare diseases, and
9 that's when these disease-specific scales can --
10 can come in.

11 Now, in the -- in the -- in the more
12 common diseases, you know, when I -- when I've
13 seen patients who have gotten disability who I did
14 not think were disabled, it was usually because
15 they had the diagnosis nailed, but the severity of
16 illness was -- was not recognized by the -- by the
17 adjudicator, I guess was assumed to be more severe
18 than it was.

19 So I think that -- that's the tricky
20 part, I think is -- is being transparent about
21 what is the severity of illness that's associated
22 with disability to eliminate your false positives,

1 but to put that into a process so it's not so
2 tricky to negotiate that you end -- end up, you
3 know, failing a lot of patients who really are
4 disabled, but they just didn't get the scale
5 measured right or something peculiar about that
6 scale in their position.

7 I think those are the kind of things
8 we have to try and work out. And you need data to
9 be able to know, you know, your sensitivity and
10 specificity of any kind of a -- any kind of a test
11 like that.

12 ACTING DEPUTY COMMISSIONER RUST:
13 Thank you.

14 MR. BARTEK: If -- if I could take the
15 liberty and just expand on that very important
16 point. Dr. Koroshetz is talking about the
17 difference between a generic assessment, which in
18 our case would be a genetic assessment, and -- and
19 severity of illness at that particular point in
20 time when eligibility might be an issue.

21 And -- and going back to the kinds of
22 scales, Dr. Koroshetz is, boy, completely accurate

1 in our experience that you can't even pretend to
2 be preparing for clinical trial if you don't have
3 scales. FDA would laugh you out of the room. And
4 those scales --

5 COMMISSIONER ASTRUE: Sadly, I've had
6 that occur.

7 MR. BARTEK: Yeah, right.

8 And those scales include not just
9 the -- the measurements that I've described, like
10 25-foot walks and pegboard tests and so forth, but
11 really, a -- a quality of life -- living, you
12 know -- a score and activities of daily living
13 score.

14 You begin with the standard forms,
15 like SF-36 for quality of living scales, but then
16 they're made specific to each of these diseases
17 in -- in the clinical experience that -- that
18 after you've seen several hundred, or in some
19 cases 10,000 patients, you know what questions to
20 ask.

21 Going back to another discussion you
22 had this morning, you're not always -- the SF-36

1 doesn't always ask the right question for this
2 particular condition. So now these wonderful
3 clinicians are refining those and adding questions
4 to the SF-36 and so forth. So these scales now
5 include that.

6 So I think they can be wonderfully
7 instructive in your task.

8 COMMISSIONER ASTRUE: Okay. Frank, do
9 you have anything you want to ask?

10 JUDGE CRISTAUDO: I -- I would like
11 to -- to ask Dr. Koroshetz. I'd like to follow up
12 on a point that he made in his testimony in
13 following up, actually, on a question that -- that
14 David asked about.

15 You mentioned, Doctor, that the
16 doctors who have to complete these forms in -- in
17 practice -- and you were in practice, it sounds
18 like, for many years -- feel pressured, there's
19 kind of a dilemma that they're faced with in -- in
20 completing those forms, and it's obviously a
21 critical issue in -- in terms of what the severity
22 is and -- and the way we make our decision on --

1 on disability.

2 And -- and I've asked this question of
3 the earlier panels, and I'm -- I'm trying to
4 determine, is there a better way for us to ask
5 the -- the medical sources for their opinion on
6 what the functional limitations are that a -- that
7 an individual has as a result of the impairments?

8 I know in many cases we send out a
9 form that lists, you know, how -- how long can
10 they sit, stand, walk and that sort of thing, and
11 there's some thinking that perhaps an alternative
12 approach would be better to ask in terms of
13 limitations imposed by the -- by the impairments.

14 And I just wonder if you had any
15 thoughts on what would be best for the medical
16 sources.

17 DR. KOROSHETZ: So I thought about
18 that and, you know, the -- the best way to solve
19 the problem would be to have -- and this is just
20 my guess now, and I have to -- maybe I'm wrong, I
21 have to think it through, but my -- my best guess
22 is that to have an objective measure of severity

1 that is really transparent is probably the easiest
2 thing to do, because -- because -- because
3 nobody -- you know, the -- the -- the -- the
4 physician wants to do the right thing, and I don't
5 think that they ever fill out the form
6 untruthfully, they put down the information that's
7 asked and -- and then the question -- and then --
8 and their issue is, okay, I put it down and let's
9 see what SSA does with it.

10 And so the -- the issues that -- if --
11 if there was a transparent scale where, you know,
12 you had followed this patient for four or
13 five years, you're going to follow them another
14 five years and you -- you scale the disability,
15 their -- their severity score, that that is a red
16 flag when the patient is getting disability and
17 their severity score is minimal to nothing.

18 I think that would probably be the
19 easiest way to go. Because it's an objective
20 thing, it's not something somebody can argue.
21 Someone has aphasia, so you put down aphasia, you
22 know, and aphasia, define inability to

1 communicate, to discuss things with coworkers,
2 and -- but the -- but you never -- but there's no
3 way in the form to actually indicate what the
4 degree of severity of the aphasia is.

5 So -- so I've seen patients who have
6 an aphasia, no question, you put in the form, they
7 have an aphasia, they had a stroke, they've
8 recovered significantly and -- but you don't
9 have -- there's no way of saying how -- how bad
10 that aphasia is. The aphasia scales which are out
11 there are not -- are not -- not asked for.

12 So no one knows -- so the diagnosis is
13 there. The patient gets disability because they
14 had aphasia after a stroke, but what no one knows
15 is the aphasia is really pretty minimal, and they
16 actually do pretty well. They can't -- you know,
17 usually in these instances, they can't perform the
18 job that they had before.

19 So a guy who was a -- you know, a CEO,
20 has a stroke, he has an aphasia, you know, he
21 could -- he could work at another level, he
22 can't -- probably can't work as a CEO anymore, but

1 that's -- you know, those are the kind of problems
2 that -- that the Social Security Administration
3 doesn't actually get a good sense of the severity
4 of each of the issues.

5 I think a scale of -- severity scale
6 in my mind would be, you know, a red flag when
7 things are not -- not -- don't seem to match,
8 severity is low. And also, you know, potentially
9 and automatic when you have a disease, as you well
10 know, and you hit this level of severity,
11 everybody knows we have 10,000 patients, you hit
12 that level of severity, everybody's -- nobody can
13 work, that would be another advantage.

14 Does that make sense?

15 JUDGE CRISTAUDO: It does.

16 Like having a GAF scale, for example,
17 but for physical impairments --

18 DR. KOROSHETZ: Yeah.

19 JUDGE CRISTAUDO: -- I think is what
20 you're saying.

21 DR. KOROSHETZ: Right. The -- the
22 issue is -- to make -- for patients -- and I guess

1 what we're getting at, and this is a tough thing
2 to swallow, is that these -- that the -- that the
3 disease is so different that to have a generic
4 scale gets -- leads to loopholes, I think. To
5 have a more specific scale for the disease is
6 probably the better way to go, but then that's an
7 incredibly complicated thing to put into place.
8 That -- that's where I see the attention.

9 JUDGE CRISTAUDO: Thank you.

10 COMMISSIONER ASTRUE: I hate to cut
11 this off because it's very helpful and -- and
12 really interesting, but it's my sad duty to do
13 that.

14 Thank you to all the panelists. It's
15 been very helpful.

16 I want to reiterate our gratitude to
17 Dr. Koroshetz and -- and all of NIH. They've just
18 been great partners on this, and I think they're
19 going to be incredibly important for us going
20 forward, and we're -- we're very grateful for the
21 support.

22 We're going to take a 15-minute break

1 now.

2 I have concluded that we were overly
3 generous with the closing remarks for the
4 panelists here, and I don't think we're going to
5 use that time. So we'll reconvene at 3:20. We'll
6 have an hour for the final panel.

7 And then we'll -- we'll -- we'll
8 restrict ourselves to no more than 10 minutes.
9 I'll still get you out of here by about 4:30.

10 So thank you very much, and we'll
11 reconvene in 15 minutes.

12 (Recess.)

13 COMMISSIONER ASTRUE: Okay. We're on
14 the home stretch here. We've got one last panel,
15 which I'm looking forward to, so I -- I'd like you
16 to interrupt the conversations again and focus our
17 attention.

18 We have four members of this panel.
19 We have Dr. R. Rodney Howell, who is President of
20 the American College of Medical Genetics
21 Foundation and Chair -- and he's also chair of
22 Secretary Leavitt's Advisory Committee on

1 Heritable Disorders and Genetic Diseases in
2 Newborns and Children.

3 We have Dr. William A. Gahl, who's
4 clinical director of the National Human Genome
5 Research Institute at the National Institutes of
6 Health with his colleague, Dr. Suzanne Hart.

7 And finally, we also have
8 Dr. Andrea Gropman, Associate Professor of
9 Pediatrics and Neurology at George Washington
10 University School of Medicine and Health Sciences,
11 attending in neurology, who's also with the
12 Children's National Medical Center.

13 Welcome.

14 Who -- who's going start? Dr. Howell,
15 are you going to start?

16 DR. HOWELL: I think that's the order.

17 COMMISSIONER ASTRUE: Okay. Great.

18 DR. HOWELL: Thank you very much,
19 Commissioner Astrue. It's my pleasure to be here.
20 And I'm going to start with some very general
21 overview comments about newborn screening.

22 The newborn screening has been an

1 active program in the United States for a little
2 over 40 years. It started with the recognition
3 that a long-known condition, that is,
4 phenylketonuria, an abnormality in the metabolism
5 of phenylalanine, could be dramatically improved
6 if infants, very early in life, were put on a
7 special diet that was deficient in phenylalanine.

8 The infants who were treated early
9 were recognized because they had a family history
10 of the condition so that they could be tested when
11 they were little babies and so forth and tried on
12 the diet.

13 It was also shown very early that if
14 you tried this new diet on older children with
15 phenylketonuria, the benefit was minimum. So that
16 you had the very striking paradigm, a dramatic
17 diet that had to be diagnosed in the newborn
18 period. And since there's no family history
19 usually in these conditions, you needed to
20 conceive of a test that could be applied to the
21 population at large.

22 At that time, the -- the question was

1 posed to Bob Guthrie, who is a Professor of
2 Pediatrics and Microbiology in Buffalo, who was
3 active in the community of persons with
4 handicapped, about could he devise a test. And he
5 come up with a very simple bacteriologic assay
6 that depended on using a dried blood spot on
7 filter paper and putting those dried blood spots
8 onto auger containing a B subtilis spore that was
9 dependent on phenylalanine. And this very simple
10 test turned out to be extremely usable in the
11 public health arena.

12 And largely because of public
13 advocacy, families with children with
14 phenylketonuria, this technology was adopted
15 rapidly throughout the country over a period of
16 several years, and this was in the early to
17 mid '60s.

18 The -- the situation stayed relatively
19 the same, technologically; that is, a dried blood
20 spot, you punch out one hole, and you test for one
21 condition. However, other conditions were added
22 that had therapies, such as maple syrup urine

1 disease, hypothyroidism, but you had this one
2 spot, one test technology. Some states actually
3 ran as many as 10 tests, all on individual little
4 holes that they punched out as tests.

5 The scene changed quite dramatically
6 in the early '90s with the widespread adoption of
7 tandem mass spectroscopy. And this technology is
8 extremely reliable and permits you, on a single
9 dried blood spot, to analyze for a number of
10 abnormal compounds, and in the areas of inherited
11 rare metabolic disease, it turns out that you can
12 really diagnose around 40 conditions in -- in a
13 matter of minutes with considerable accuracy.

14 And since that time, there's been a
15 considerable increase in the number of tests that
16 are done in newborn infants in this country. At
17 the current time, the national recommendations
18 from both the advisory committee that I chair and
19 also from the American College of Medical Genetics
20 is that there are 29 conditions that are
21 appropriate for newborn screening in the so-called
22 core panel.

1 These conditions all share the common
2 denominators: One, they're all very serious, they
3 all have treatments that, if begun earlier, are
4 beneficial. And the other thing is they're not
5 evident at birth; in other words, the baby looks
6 normal, so there's no sign that would say,
7 goodness, this baby has this problem.

8 Now, state newborn screening --
9 newborn screening programs are, in all instances,
10 operated by the states. And so what is screened
11 in Virginia, is decided in Virginia, et cetera.
12 But it's very been interesting to me from a
13 national level to look at what's happened since
14 these recommendations have gone down in the past
15 few years. Because as we sit here today,
16 90 percent of all the babies born in the
17 United States are getting that -- that core panel
18 done. And so the states have actually quite
19 rapidly adopted that panel.

20 It'd be appropriate to comment at this
21 time is that there are 4.1 million babies born
22 each year in the United States, and since they are

1 all getting genetic testing and many or most of
2 them, as many as 30 conditions, newborn screening
3 for genetic diseases is by far the most common use
4 of genetic testing that's done today in this
5 country.

6 I think these conditions are
7 particularly interesting for a variety of reasons.
8 One is that people with these conditions are born
9 with what I would call a permanent abnormality or
10 disability. In other words, they lack a protein
11 or they lack an enzyme. However, if detected very
12 early, they can commonly lead an essentially
13 normal life.

14 And recent -- the recent NIH panel on
15 the treatment of phenylketonuria had a college
16 student as a member of the panel who herself had
17 PKU. An untreated PKU, the average intelligence
18 is under 25, so the benefits are enormous.

19 However, there are many programs
20 currently being screened for in the country that
21 are not, shall I say, "curative." For example,
22 most states currently screen for cystic fibrosis

1 and although the outcomes are improved, children
2 with cystic fibrosis persist with disability, as
3 you know, although they do much better if
4 diagnosed early, et cetera.

5 The other widespread screening
6 condition today that falls in a similar category
7 is all states are either implementing or currently
8 screening for hearing defects.

9 And although you can benefit these
10 children by early identification of the defect,
11 they commonly will need either cochlear
12 implantation or assistive devices in order to
13 hear. So they will need long-term support and
14 help.

15 And I'd like to just comment about the
16 fact that there are dramatic new technologies on
17 the scene. I've talked about tandem mass spec and
18 so forth. But currently under look -- under
19 investigation are some microfluidics which use
20 extraordinarily small samples and can screen for a
21 large number of -- of areas.

22 There are new diseases that are being

1 screened for that have therapies coming up. Two
2 lysosomal storage diseases that are currently in
3 pilot studies in the United States for newborn
4 screening are Krabbe disease, a historically
5 rapidly fatal disease that, if identified in early
6 infancy and treated with a bone marrow transplant,
7 has dramatic improvement. These children are not
8 cured, they still have disability, but they're
9 dramatically improved and they survive.

10 The other is Pompe disease, a
11 lysosomal storage disease that, in its most severe
12 form, causes early infant death, is under
13 treatment with enzyme replacement therapy, which
14 is being introduced for a number of conditions, as
15 the Commissioner certainly knows, having been
16 seminally involved in this area himself in the
17 past.

18 I think that the -- the -- the point
19 is, is that the technology that's being used in
20 newborn screening is highly accurate. One
21 commonly talks about, you know, you have a certain
22 number of false positives, and I would say that

1 the term "false positive" is not the right term,
2 because false positive suggests that you have an
3 error in laboratory diagnosis or you have
4 something, but we have physiologic variance with
5 chemical changes in the baby. And so a test may
6 not be within your normal limit and when repeated,
7 is normal.

8 But I would call we need to come up
9 with a better term. There's variation and so
10 forth, but the technology that's being used is
11 extremely reliable and very accurate.

12 And I think one of the major things
13 that's come out of the newborn screening needs in
14 recent years has been the fact that once we
15 identify an infant -- and about 1 in 1,000 babies
16 born in this country will screen positive for one
17 of these conditions, so it's a substantial number
18 of -- of infants. Although they're individually
19 rare when you put them together, it's a
20 substantial number.

21 We need to develop a system to put all
22 of those into a long-term follow-up program to see

1 exactly how they're doing with treatment, what's
2 happening to them, and also make available these
3 children and young adults for new -- new therapies
4 and new technology.

5 And this is one of the areas where the
6 National Institutes of Health is looking very
7 carefully at how to institute -- is to implement a
8 program that would study the -- all the babies
9 that are born positive in this country.

10 So those are my opening remarks.

11 Thank you very much.

12 COMMISSIONER ASTRUE: As has become
13 traditional today, we're going to hold off our
14 questions until the end and then -- because we
15 figure there'll be overlap in response. Okay?

16 DR. GAHL: Okay. Thank you.

17 Well, medical technology can bring us
18 to a diagnosis and can often bring us objectivity
19 to that diagnosis, and sometimes the diagnosis can
20 bring us a prognosis as well.

21 So Dr. Hart is going to talk about
22 some specifics of those medical technologies, and

1 then I'm going to offer a perspective after --
2 after she speaks for just a few moments. And
3 Dr. Hart has wide experience in molecular and
4 biochemical diagnostics, so she's going to tell us
5 a few things about those specifics.

6 DR. HART: The Task Force on Genetic
7 Testing, convened by the National Institutes of
8 Health, Department of Energy Joint Working Group
9 on the Ethical, Legal and Social Implications of
10 Human Genetic Research, defined a genetic test as:

11 The analysis of human DNA, RNA,
12 chromosomes, proteins and certain metabolites in
13 order to detect heritable disease-related
14 genotypes, mutations, phenotypes or karyotypes for
15 clinical purposes. Such purposes include
16 predicting risk of disease, identifying carriers,
17 establishing prenatal and clinical diagnosis or
18 prognosis. Prenatal, newborn and carrier
19 screening, as well as testing in high-risk
20 families are included.

21 Tests for metabolites are covered only
22 when they are undertaken with high probability

1 that an excess or a deficiency of the metabolite
2 indicates the presence of heritable mutations in
3 single genes.

4 Tests conducted purely for research
5 are excluded from the definition, as are tests for
6 somatic, as opposed to heritable mutations and
7 testing for forensic purposes.

8 Individuals who undergo genetic
9 testing fall into two main categories: One,
10 symptomatic individuals are those in whom a
11 specific clinical feature or phenotype suggests a
12 genetic disorder; or, two, individuals with a
13 family history of a genetic condition.

14 Current genetic testing methodologies
15 are broadly categorized into biochemical,
16 molecular and cytogenetic techniques.

17 This testimony will focus on
18 biochemical and molecular techniques.

19 Existing molecular genetic testing
20 includes targeted mutation analysis, for example,
21 by targeted sequencing, allele specific
22 oligonucleotide analysis, et cetera, whole gene

1 sequencing, typically covering coding regions and
2 exon/intron splice junctions, determination of
3 number of repeats for trinucleotide repeat
4 disorders, deletion/duplication analysis, and
5 determination of epigenetic changes such as
6 methylation status.

7 Detection rates vary based upon the
8 gene and the methodology used. As sequencing
9 costs have fallen, full sequencing has become more
10 common. This will likely result in higher
11 detection rates compared to targeted analysis.

12 In the future, array technology will
13 be more frequently employed in the molecular
14 diagnostic laboratory. There are two main areas
15 where this technology will be helpful. First, for
16 large genes, array technology provides a
17 cost-effective and time-saving platform for
18 mutation detection.

19 For example, the dystrophin gene,
20 which is mutated in the two allelic disorders,
21 Duchenne muscular dystrophy, DMD, and Becker
22 muscular dystrophy, spans more than 2 million

1 basepairs of DNA.

2 More than 500 deletions,
3 80 duplications and almost 1,000 single base
4 changes have been described in this gene. Two
5 arrays have been developed to aid in genetic
6 testing for these muscular dystrophies. The first
7 array detects deletions and duplications of the
8 DMD gene, which account for approximately 70 to
9 75 percent of mutations in DMD and 90 to
10 95 percent of mutations in Becker muscular
11 dystrophy.

12 The second array is a sequencing array
13 that allows the whole DMD gene to be interrogated
14 in a single assay.

15 Another use of array technology will
16 be the evaluation of multiple genes
17 simultaneously. The advantage of this technology
18 can be illustrated with X-linked mental
19 retardation.

20 Currently, 63 genes on the
21 X chromosome are known to be associated with
22 syndromic X-linked mental retardation. This is

1 mental retardation associated with other
2 phenotypic features.

3 An array is under development that
4 would allow analysis of 34 genes on the
5 X chromosome concurrently. At the present time,
6 such analysis would have to be conducted
7 sequentially at a significantly higher cost and
8 turnaround time.

9 Biochemical testing for rare diseases
10 involves standard blood chemistry, hematological
11 and immunological studies, as well as specialized
12 tests for rare metabolic disorders. These include
13 amino acid and organic acid analysis, enzyme
14 assays, antibody tests, specific small molecule
15 assays and profiles of abnormal metabolites.

16 As Dr. Howell has mentioned, tandem
17 mass spectrometry is revolutionizing newborn
18 screening, and this method has the potential to
19 improve the biochemical diagnosis of symptomatic
20 patients as well as newborns.

21 The technique can allow faster and
22 perhaps more definitive diagnosis of known

1 diseases and can reveal new disorders by detecting
2 small molecules that signal disease.

3 Ultimately, the best testing
4 methodology varies with the disorder or gene
5 involved and may also depend upon ethnicity. For
6 example, carrier testing for Tay Sachs disease, in
7 the absence of family history, is best performed
8 by molecular means for Ashkenazi Jewish
9 individuals, but by biochemical methods for
10 nonAshkenazi Jewish individuals.

11 In some cases, combined testing may be
12 the most appropriate approach.

13 DR. GAHL: I would say that these
14 medical technologies get us to a diagnosis, and
15 sometimes that diagnosis is incredibly meaningful
16 in itself; that is to say, that everyone who has
17 the diagnosis will have a disability. And
18 examples of that could be Progeria, which is a
19 premature aging syndrome in which the patients die
20 at about 13 years of age. Or a Fragile X or
21 Tay Sachs disease, which is a lysosomal storage
22 disorder, or other lysosomal storage disorders,

1 adrenoleukodystrophy, for example,
2 Usher's syndrome, which will predict deafness, or
3 albinism, which will predict blindness.

4 But then there are other disorders in
5 which the diagnosis does not itself predict
6 disability, and you need to know something else
7 about it. For example, in Huntington's disease,
8 you know eventually there's going to be problems,
9 but you may not -- if you carry the gene, you may
10 not have the problem at age 15.

11 And the same goes for some of the late
12 onset myopathies like Hereditary Inclusion Body
13 Myopathy and even Fabry disease, which has later
14 onset to these disorders.

15 Lists of these types of disorders, the
16 ones that by themselves are going to give a
17 problem immediately and the ones that may later
18 can be constructed with some reasonable
19 objectivity because of medical technology and
20 expertise. And that's the second point that I'll
21 come to in a little bit.

22 But most patients who come with a rare

1 disease diagnosis will eventually require medical
2 assistance. The issue is at what time is the
3 diagnosis being made, what time are they being
4 evaluated. If they come to medical diagnosis and
5 use medical technology at the time when they have
6 symptoms, they probably already need intervention.

7 But sometimes the genetic diagnosis is
8 made before that, and it's only later that they'll
9 need to -- to be declared disabled and -- and
10 receive assistance.

11 Furthermore, there are different rates
12 of progression of these disorders, and some of the
13 disorders are even static.

14 Well, I think that the new
15 technologies that will come about will not change
16 this basic paradigm incredibly much. In other
17 words, the diagnosis will always take us so far,
18 but never so that we can completely predict for
19 all of these rare diseases that disability is
20 required.

21 The bulk of the decisions regarding
22 these allowances also require input from health

1 care professionals, experts in the field, and
2 fortunately, for many of the rarer diseases,
3 there's a -- for the metabolic disorders, there's
4 a society for inherited metabolic disorders, and
5 that society has those experts. And I think that
6 as a group, the SIMD would be willing to offer its
7 expertise in the adjudicatory process, along with
8 other health care professionals.

9 So they could do this on an individual
10 label -- basis, but they could also offer advice
11 a priori by creating some of those lists of the
12 disorders which are certain to require disability
13 allowance and those that may or may not, based
14 upon particular symptoms.

15 Thank you.

16 DR. GROPMAN: First I'd like to say
17 it's my pleasure to be here today, and I commend
18 the Commissioner and his committee for convening
19 this hearing.

20 As you've heard from my colleagues,
21 our knowledge of inborn errors of metabolism and
22 our screening in recent years has expanded at an

1 impressive rate as a result of research in
2 laboratories and clinic -- clinical trials around
3 the world.

4 Rapid diagnosis and, when possible in
5 treatable conditions, metabolic rescue, is
6 mandatory to improve neurologic outcome as the
7 majority of these conditions that were discussed
8 impair the neural access in some way.

9 In addition, advances in technology
10 have made early diagnosis feasible, and with this
11 recognition, the outcomes of treatment better.

12 As new treatments are emerging, they
13 will impact on outcomes. One consequence that
14 you've heard of early screening and treatment is
15 that children with these diseases that were once
16 considered uniformly fatal are now surviving into
17 adulthood; some as healthy productive citizens,
18 others with significant challenges ahead of them.

19 Despite these advances, however, there
20 continue to be barriers that exist leading to
21 diagnosis and provision of services to individuals
22 with these conditions.

1 Many of these conditions are expected
2 to worsen over time. And although physical
3 disability may be expected, the impact on
4 neurologic function, cognitive changes and mental
5 function may be less clear.

6 Likewise, the discovery of new
7 variants of previously described metabolic
8 diseases has added further to the complexity of
9 diagnosis of inherited metabolic disorders.

10 Late onset in adult variants of
11 diseases formerly thought to occur only in infancy
12 or childhood are being diagnosed not infrequently.
13 And often, the clinical features of the disease in
14 older individuals is markedly different from that
15 seen in infancy.

16 As a child neurologist and geneticist,
17 I have firsthand experience in the problems of
18 getting to diagnosis and being able to determine
19 disability.

20 Neuroimaging of inborn errors of
21 metabolism has begun to paralyze advances in other
22 technologies. As the central nervous system

1 effects of some congenital and acquired infections
2 are recognized by characteristic distributions of
3 abnormalities on neuroimaging studies, likewise,
4 specific imaging findings may, in some cases,
5 strongly suggest a metabolic disorder or inborn
6 error of metabolism as the etiology of the
7 patient's neurologic symptoms or disability.

8 In fact, large collaborative research
9 studies, predominantly in adult degenerative
10 disorders, have convened to look for biomarkers
11 that may actually predict the onset severity as
12 well as response to treatment in a number of
13 conditions which could impact the determination
14 for disability.

15 While in other cases the imaging
16 features of metabolic disorders may mimic that due
17 to other causes, there are certain well-described
18 imaging patterns that strongly support the
19 diagnosis of a metabolic disorder.

20 These patterns, in turn, may depend
21 upon the age and stage of the disease at the onset
22 of symptoms, and furthermore, cell type and

1 regional selective vulnerability to metabolic
2 toxins and ischemia are responsible for the
3 differing imaging patterns that are seen.

4 The differential diagnosis of a
5 metabolic disorder is thus narrowed, depending on
6 whether the findings affect primarily the gray or
7 white matter, both, and there is typically an
8 imaging signature that may be recognized, such as
9 an altered signal in the basal ganglia in glutaric
10 aciduria Type I, the parietal occipital changes
11 seen in MELAS and other mitochondrial disorders,
12 absence of the creatine peak on magnetic resonance
13 imaging spectroscopy seen in creatine deficiency
14 disorders, or an increase in N-acetylaspartate, a
15 putative neuronal marker which is seen in
16 Canavan's disease.

17 Likewise, a common imaging finding in
18 patients with changes in mental status may be that
19 of edema caused by vasogenic or cell swelling and
20 may accompany stroke, infection or metabolic
21 disorder.

22 The location of the swelling, for

1 example, being in white matter, brainstem and
2 cerebellum, would invoke the diagnosis of maple
3 syrup urine disease, for example, and not hypoxic
4 ischemic injury in an infant presenting with
5 changes in mental status and edema.

6 Certain organic acidemias, such as
7 propionic or methylmalonic have a predilection for
8 causing signal changes on MRI in white matter and
9 basal ganglia, and the urea cycles classically
10 show elevations of glutamine, as can be measured
11 noninvasively by magnetic resonance spectroscopy.

12 Because inborn errors in metabolism
13 can present in the neonatal period with neurologic
14 distress, metabolic acidosis and multisystem
15 involvement, similar features encountered in
16 hypoxic-ischemic encephalopathy, an underlying
17 metabolic disorder may be undiagnosed, unless the
18 clinician maintains a high index of suspicion and
19 performs appropriate diagnostic testing.

20 This certainly will impact the ability
21 to recognize and provide needed services for these
22 individuals.

1 So, as I've indicated, magnetic
2 resonance imaging has emerged as a powerful tool
3 in the study of both normal and abnormal brain
4 structure, function and biochemistry.

5 It is a nonionizing and noninvasive
6 technique and most, if not all, medical centers
7 have access to one or several scanners, and
8 imaging of pediatric patients, including our
9 premature infants, is now quite routine.

10 While skillful assessment of the
11 results is required, there are several references
12 to assist with this process. MRI can allow the
13 differentiation of cell bodies or gray matter from
14 the connecting fibers, or the axons, which appear
15 in the white matter.

16 And with a technique called magnetic
17 resonance spectroscopy, or MRS, brain biochemistry
18 can be sampled in a noninvasive way, and this
19 information taken with the clinical features to
20 aid with diagnosis.

21 In addition, newer modalities of
22 imaging are moving from the research scanners into

1 more widespread clinical use. For example, white
2 matter microstructure can be studied using a
3 technique called diffusion tensor imaging, which
4 may allow abnormal white matter to be visualized
5 prior to seeing it on conventional MRI.

6 And magnetic resonance spectroscopy,
7 to study brain metabolism, can allow for
8 delineation of regional metabolic changes as a
9 result of disease progression or, as therapeutic
10 interventions are introduced, can be used to
11 measure outcome.

12 Neuroimaging studies may be indicated
13 in a majority of patients with mental retardation,
14 developmental delay, acute encephalopathy, loss of
15 skills or cognitive impairments of unknown
16 etiology.

17 Even in conditions that are due to
18 known causes, the MRI can be used to assess the
19 degree of involvement, which may help contribute
20 to disability determination.

21 The most common imaging techniques
22 used currently in the diagnostic evaluation of

1 individuals with mental retardation or
2 disabilities are CAT scan, MRI and magnetic
3 resonance spectroscopy. However, given the unique
4 gray/white matter differentiation, MRI is probably
5 the best imaging test in a diagnostic workup.

6 Some conditions in which neuroimaging
7 procedures, and specifically MRI, have proven to
8 be useful include the following cases, which I'll
9 discuss:

10 Neuronal migration disorders, brain
11 malformations or hypoplasias, intrauterine
12 infections, disorders due to birth injury,
13 vascular disorders, white matter diseases,
14 including demyelinating disorders such as ALD,
15 multiple sclerosis, metachromatic leukodystrophy,
16 and Canavan disease; and other metabolic
17 conditions, organic acidemias, mitochondrial
18 disorders, urea cycle disorders.

19 Because of the huge array of available
20 molecular and biochemical tests, the information
21 from neuroimaging may help focus the search.

22 The technologies of today that are

1 useful within the MRI scan package itself include
2 FLAIR imaging to view white matter,
3 MR spectroscopy and diffusion tensor imaging.

4 And in the future, improvements of
5 these methods with higher field scanners and other
6 types of metabolic imaging will certainly improve
7 early diagnosis.

8 Therefore, the delivery of SSI and SSA
9 services to individuals with rare disorders will
10 depend upon early diagnosis, understanding of the
11 diagnosis -- of the disorders and their effects on
12 function and independent living, and understanding
13 the limitations may not be static, but, in some
14 conditions, are expected to be progressive over
15 time.

16 The major challenge and question,
17 therefore, is to how to use and integrate these
18 technologies in a way that will impact disability
19 determination with the goal to establish an
20 accurate and valid system that benefits those
21 patients in needs of services.

22 I believe I speak also on behalf of my

1 colleagues that, as a group, neurologists and
2 geneticists are available to engage in such
3 dialogue to address this, as we appreciate the
4 importance and complexity of this disability
5 determination process.

6 Thank you.

7 COMMISSIONER ASTRUE: Thank you.

8 Thank you. I've got a couple questions.

9 We, of course, because of our mission,
10 have to ask very different questions than what
11 doctors trying to treat disease or companies
12 trying to provide therapies ask.

13 So we're -- we're very focused on
14 trying to figure out how markers correlate with
15 functionality.

16 So I wanted to ask first on -- on
17 genetic testing. A lot of the lysosomal storage
18 diseases, for instance, have a wide range of
19 severity. Pompe disease, you can have children
20 dead at the age of 2, but I've also had the
21 experience of talking to a woman in her 30s for
22 15 minutes before she disclosed she had

1 Pompe disease, and I had no clue.

2 So there -- there's a range, and
3 something like Hunter's, somewhere, 5, 6, it'll
4 split pretty evenly between those kids that have a
5 pretty good shot at not only survival, but living
6 pretty normal lives, with some of the new
7 therapies and those where the disease attacks the
8 central nervous system, and they're looking at a
9 horrible death, you know, in a five- to eight-year
10 period.

11 To what extent are we able now,
12 through genetic testing, to sort out within those
13 diseases that have a range of severity, can we
14 tell genetically at birth what the likely course
15 is? For instance, with a Hunter's child, which
16 one is likely to have seen as complications or not
17 or -- or is that we're just not there yet in terms
18 of being able to use this -- these data?

19 DR. GAHL: I'm -- I'm going to let
20 Andrea answer in a minute, but I don't think we're
21 really there yet with respect to genetic testing.
22 It turns out that there's relatively poor, or

1 let's say incomplete genotype, phenotype
2 correlations for most of these disorders. And
3 that may fill out over the next, let's say, decade
4 or two, because the clinical characteristics are
5 then associated with certain mutations.

6 But there's a problem here, maybe not
7 quite so much for Hunter's as for some of the
8 others that are autosomal recessive, you have
9 compound heterozygotes -- so, in other words, two
10 different mutations -- and you can't really
11 tell -- you can tell that there's a severity
12 associated with one mutation, but not so well when
13 there are two different mutations, essentially.
14 So -- so, you know -- so -- so that's -- that's an
15 issue.

16 It turns out that some of our best
17 phenotype correlations are with biochemical
18 measurements. So the amount of residual enzyme
19 activity does correlate pretty well with the
20 severity of disorders, and that goes way back.

21 So we should perhaps move ourselves a
22 little bit away from the molecular and towards the

1 biochemical, which essentially measures all sins.
2 You know, it's the final measure of how much
3 residual competency you have for that enzyme
4 pathway.

5 I'd also say that differential tissue
6 expression can be a problem here. In other words,
7 you might have a particular mutation in a gene
8 such that there's a reasonably normal amount of
9 enzyme activity within the liver or the kidney or
10 some such, but not so much in the brain, and that
11 can account for some of -- some of the
12 differences, too. And we're not very good at
13 that, because we don't get those organ or tissues
14 to measure the enzyme in.

15 COMMISSIONER ASTRUE: What -- what
16 percentage of diseases? Is it just a question of
17 brute force as opposed to sort of an intellectual
18 issue that needs to be resolved? Do you have any
19 sense of that?

20 DR. GAHL: What -- what percentage of
21 disease is what?

22 COMMISSIONER ASTRUE: Would -- would

1 we be able to perhaps sort out the severity of the
2 disease from gene testing as opposed to waiting to
3 see what happens? Is it -- is -- is it possible,
4 do you think, in 5 percent, 10 percent if we do
5 the work? Is it higher than that?

6 DR. GAHL: I -- I think it's higher
7 than that, actually. It -- it has to do with
8 accrual of patients and performing clinical
9 characterization at the same time as doing the
10 molecular diagnostics. So to a certain extent,
11 this is a matter of time and -- and money.

12 You could probably do it now for half
13 or more of the disorders. It's just that, you
14 know, there aren't grants for that, there's no
15 real support for that.

16 COMMISSIONER ASTRUE: Right.

17 DR. GAHL: Companies are coming close
18 on occasion to doing that because it's sort of in
19 their best interests.

20 COMMISSIONER ASTRUE: Okay.

21 DR. HOWELL: Let me comment. I
22 certainly agree with everything that Bill said and

1 the -- I mentioned the fact that the NIH is very
2 interested in following up on the newborn kids,
3 and one of the things that will be looked at
4 keenly is looking at all the markers, looking at
5 enzyme activity and correlating that with
6 long-term follow-up and see how that works out.

7 Now, the other thing is that
8 New York State has had to deal with this in spades
9 in their screening program for Krabbe disease.
10 They have mandated screening in the state for
11 Krabbe disease and, like Pompe disease, there's
12 different forms of Krabbe. Some are acutely ill
13 as infants and others have a later onset.

14 And so what they've done, and I think
15 they worked very hard on this, is to, once the
16 patient is identified as having an enzyme
17 deficiency, they basically then have a whole
18 variety of other tests that they put into play,
19 such as nerve conduction velocity, there's a
20 variety of other things, and try to make a
21 judgment about, you know, what's going to be the
22 time frame of that patient. And I think those

1 will clearly be informative when -- when they --
2 when they finish their studies over this time.

3 But they've had to deal with -- and
4 Pompe, of course, is a perfect example. The --
5 Bill mentioned the professional groups are looking
6 at paradigms in the diagnosis and treatment and
7 the -- the college has been working with SIMD on a
8 paradigm in the diagnostic follow-up of patients
9 with Pompe disease. Because once you get a
10 patient with Pompe disease, you're going to have
11 to make some decisions fairly early, because some
12 of the patients will have massive cardiomegaly and
13 others will be asymptomatic.

14 And I think doing genotype analyses
15 will be helpful, but the correlations have not
16 been very good. But enzyme activity, according to
17 the Duke group, has been fairly predictive.

18 COMMISSIONER ASTRUE: Okay. I've got
19 one more question, and then I'll turn it over to
20 my colleagues. I'll start with Dr. Gropman, but I
21 don't, you know, want to stop anyone else from
22 jumping in.

1 And this is a little bit out of our
2 mandate today, but that's okay. You're all here,
3 so I'm just going to ask what I want, so --

4 We -- we have -- one of the -- we have
5 a lot of claimants in certain categories in the
6 neurological diseases, and it can often be very
7 hard for us to sort out for these degenerative
8 diseases sort of the break point where they fit
9 the standards that Congress has told us that
10 we -- we have to apply.

11 And so one of the things we first
12 started to talk to NIH about was multiple
13 sclerosis and looking at whether we could look at
14 a measure like neuronal scarring in the brain and
15 correlate that with functionality.

16 And one of the things that came back
17 at -- at some point was, well, the brain as a
18 whole, that might be difficult, because depending
19 on -- on where the scarring is, that might make a
20 big difference in terms of functionality.

21 Do you have any advice to us if
22 we're -- we're -- I mean, one -- one of the things

1 that would be a blessing for us and for -- for
2 claimants if we could sort it out would be some
3 efficient ways to handle these types of
4 neurological diseases.

5 Do you think that we're likely to find
6 certain areas of the brain, if we focused on sort
7 of subsets, not the brain as a whole, and could
8 track damage like neuronal scarring in MS, are
9 there certain areas of the brain that perhaps we
10 ought to be more focused on and we're more likely
11 to be able to prove some sort of correlation to
12 functionality?

13 DR. GROPMAN: I think it's more
14 complex than that. And basically --

15 COMMISSIONER ASTRUE: I was afraid of
16 that.

17 DR. GROPMAN: Yeah. Unfortunately, it
18 has to be on a case-by-case basis, because with
19 some of these metabolic conditions, I didn't
20 mention that in addition to the enzyme defect,
21 there may be other compounding factors, such as a
22 patient may have seizures, for example, which adds

1 further disability.

2 You know, it -- certainly the cortex
3 being affected will impair cognitive function, but
4 that's not to say that having a lesion in the
5 brain stem which affects basic functions such as
6 swallowing and -- and the like isn't significant
7 as well.

8 So it -- and, you know, certainly a
9 cerebellar lesion can affect the ability to walk,
10 you know, without falling.

11 So I think, you know, it will have to
12 be a case-by-case basis. I -- I believe that we
13 do have limitations with imaging.

14 We certainly have a number of impaired
15 children who have, at least structurally, normal
16 MRI scans, so we know that there's more to be told
17 than just, you know, the brain looks fine, but
18 obviously it's not functioning. Well, we're not
19 able to image on the level of synapses, for
20 example, in kids with autism and, you know, we're
21 not there yet.

22 So I think it's more complex than

1 that. You may -- for each disease group, may be
2 able to categorize disability some, but I think
3 just taken in general, it's much more complex than
4 that.

5 COMMISSIONER ASTRUE: Okay.

6 DR. GROFT: A -- a couple questions.
7 I guess first, how -- how many inherited metabolic
8 disorders would fall under the umbrella of the
9 SIMD, the group that would look at those diseases?
10 Do we have any -- any approximate numbers?

11 DR. GAHL: Well, Andrea, you -- you
12 can help me out on this, but probably in the range
13 of 100 or so. There are 225 or 250 members --
14 well, actually, there are 450 members of the
15 society, but in terms of actual physicians taking
16 care of patients, there are probably about 225 or
17 so. And they overlap some. So I'd say maybe 100,
18 150 different diseases.

19 DR. GROFT: And -- and so the society
20 members that you have probably would have pretty
21 good knowledge of most of these disorders that --
22 that they really could -- you could bring them

1 together without too much difficulty to -- to
2 address the issue of, you know, the level of
3 functioning and --

4 DR. GAHL: Yes. I think the SIMD
5 recognizes its service role to the community and
6 to the country. And I think that they would be
7 anxious to offer information on this and to assist
8 in something that the Social Security
9 Administration wants -- wants to establish.

10 Basically, the physicians in the SIMD
11 take care of these patients and deal with these
12 issues all the time. They actually write letters
13 for their patients to the Social Security
14 Administration.

15 So they're very interested in the --
16 in the care of -- of the individuals with
17 metabolic diseases. And there's a great lack of
18 understanding on the parts of -- of people all
19 over the country concerning the severity and the
20 functional effects of these rare disorders.

21 DR. GROFT: Okay. Thanks.

22 ACTING DEPUTY COMMISSIONER RUST: Sort

1 of following up on the line of questioning the
2 Commissioner started with and also the last panel,
3 they began the discussion to -- or surfaced the
4 idea that there are scales that researchers and
5 physicians use to measure severity in many of
6 these disease categories.

7 That -- that interests me a great deal
8 because one of the things we would like to do is
9 begin to find more objective measures of the --
10 the severity of -- and the disabling aspects of --
11 of disease.

12 Can you comment on that or add
13 anything to that discussion?

14 DR. GROPMAN: Okay. I think there are
15 some limitations with the scales, one being
16 certainly many of them don't apply to pediatric
17 neurologic patients. I'll take MS as an example.

18 So MS in children can be very
19 different than it is in adults, and those scales
20 may not apply. Even just to take diagnostic
21 scales, the criteria for NF1, which, you know,
22 were ingrained in all of us going through medical

1 school and training, really, you're hard-pressed
2 to make a diagnosis in a child under, you know,
3 puberty age using just, you know, the -- the
4 diagnostic criteria from the NF1 collaborative
5 scales.

6 So I think -- and -- and child
7 neurology society is keenly aware of the fact that
8 we don't have scales to the level that our adult
9 neurologic colleagues do and are actively working
10 on it.

11 I know, you know, for cerebral palsy,
12 there are some -- there are scales. There's a
13 book called, you know, Book of Scales, but whether
14 they're all applicable, you know, is another
15 issue. So I think -- and the same would go for
16 some of these metabolic conditions, because even
17 children with the same metabolic disorder may
18 present different manifestations. One child may
19 be still ambulatory with predominantly cognitive
20 effects, whereas another child with the same
21 disorder may be in a wheelchair, for example.
22 So --

1 COMMISSIONER ASTRUE: If -- if I could
2 cut in, I don't want discourage, but I just
3 have -- want to ask a specific question. For the
4 childhood MS cases, are those always going to end
5 up being severe cases as adults, or do you get
6 cases where you have onset very early and it's
7 very intermittent, the way sometimes some of the
8 adult cases are?

9 DR. GROPMAN: I think the story's just
10 unraveling with childhood MS. You know, certainly
11 making the diagnose -- the definitive diagnosis of
12 MS in a child versus its close cousin, or
13 acute demyelinating encephalomyelopathy is -- is a
14 problem. You know, a child who presents with one
15 demyelinating event, you know, what is it about
16 their event or them that will predict whether they
17 have MS or -- or they don't have MS.

18 So I think that's -- that's unraveling
19 now. There is a working group within child
20 neurology society addressing those issues.

21 DR. GAHL: I'd like to sort of
22 interject, too, about the point about geneticists.

1 We see patients who often have multisystemic
2 disorders, so that there isn't really so much a
3 scale with respect to one particular symptom, it's
4 a question of whether, for example, a cystinosis
5 patients -- patient has just renal disease or also
6 has muscle disease, swallowing difficulties,
7 pulmonary disease or central nervous system
8 problems or diabetes because those are
9 complications, and they're fairly discrete as
10 opposed to scaled. You know, they've got it as a
11 complication or not.

12 When I need functional scales, I
13 generally get it from the rehabilitation medicine
14 folks, who are very good at -- at this and -- and
15 measure function in terms of range of motion or
16 abilities to do certain activities of daily
17 living, and they're the best for me when I need a
18 consultation.

19 COMMISSIONER ASTRUE: Frank.

20 JUDGE CRISTAUDO: I would like to ask
21 the question that I've asked the earlier Panels.
22 Obviously a very distinguished panel, and it would

1 be very helpful.

2 Obviously, it's critical for us to
3 have the relevant information about the
4 individuals' impairments and so on.

5 Is there anything that we could be
6 doing in -- in terms of when we ask for
7 information from medical sources to make it
8 easiest for the medical sources to provide the
9 information to us?

10 If anyone has any comments on that.

11 DR. HOWELL: I don't have any comments
12 on it.

13 DR. GAHL: Are -- are you -- are you
14 sort of asking about Freedom of Information or are
15 you asking for specific information to be
16 garnered?

17 JUDGE CRISTAUDO: See, we're -- we're
18 trying to make determinations whether or not the
19 claimants' impairments meet a particular list of
20 impairment in our -- in our listing of
21 impairments, or we're trying to assess function,
22 and some of the earlier questions have referenced

1 that.

2 So the question really is, is we have
3 some difficulty sometimes having medical sources
4 provide information to us about -- about their
5 patients, essentially. So what -- what I'm
6 looking for is, is there an easier way for -- for
7 medical sources to provide that information to us?
8 Should we be asking the questions differently? Is
9 there anything else we can do to facilitate that
10 process?

11 DR. HOWELL: I think Bill has made an
12 important comment, is that the spectrum of
13 conditions we work with is such -- for example, in
14 Pompe disease, you need a cardiology evaluation,
15 the cardiac problem, et cetera. Another, you
16 have, in some of the others, you may have a
17 neurologic problem completely, you have a
18 neuromuscular.

19 So I think a task force, shall we say,
20 at one of the referral centers is I think the
21 thing that's going to be helpful, and -- and
22 you're going to need all of that.

1 DR. GAHL: I -- I think -- I may -- I
2 think --

3 COMMISSIONER ASTRUE: Not to jump on
4 the -- the Panel's answer. I mean, I think one of
5 the things that's going to be important for us as
6 we start listing the -- these diseases is to flag
7 for the examiners the type of tests they should be
8 running, because they don't know, because they
9 don't have familiarity with the disease.

10 You know, they could see a description
11 of Fabry disease and not realize that you should
12 be looking primarily probably at the heart and the
13 kidney. And it will -- could be an awful lot more
14 efficient for us if -- if we have instructions to
15 our examiners where, you know, that's sort of
16 flagged out for them instead of trying to get
17 them, their general doc to find an expert who can
18 give them that answer, and a lot of times we have
19 a lot of difficulty getting expert consultants in
20 these -- in the rare disease areas.

21 DR. HOWELL: One of the things I think
22 can be very helpful along that line are the

1 practice guidelines that are being developed for
2 virtually all of these conditions that basically
3 will flag, you know, what other systems that
4 become involved. And they currently are in the
5 process of being developed for many of these.
6 They've been developed for Pompe and Fabry and
7 Krabbe, but they're coming along for the others.

8 COMMISSIONER ASTRUE: And who
9 generally has the lead in those practices?

10 DR. HOWELL: The American College of
11 Medical Genetics. Again, a lot of overlap with
12 SIMD because we have -- I'm a former president of
13 SIMD, but I currently am now with ACMG, so there's
14 a lot of overlap between the groups, but they work
15 together on those. They're usually published
16 under ACMG Guidelines.

17 DR. GAHL: And I do think that for a
18 particular diagnosis, you could probably, a
19 priori, get SIMD members who are experts in that
20 to create a list for that particular disease
21 for -- for -- for you to request from the
22 physician. And you could probably do it for many,

1 many disorders; not complete, but -- but many, and
2 that could serve as sort of the first measure or
3 guide for what you need to make that assessment.
4 And I think the SIMD and other experts would be
5 willing to do that.

6 COMMISSIONER ASTRUE: Thank you. Go
7 ahead.

8 ACTING DEPUTY COMMISSIONER RUST: If I
9 can sort of take another tact on -- on
10 Judge Cristaudo's question.

11 Our examiners are looking -- the --
12 the treating physician is looking at the patient
13 one way, and our examiners are looking at the
14 patient another way. And we sometimes have a
15 sense that we don't ask for the right material or
16 the -- the physician doesn't send us the material
17 we need to -- to -- that there's a -- there's a
18 gap in terms of understanding or a -- a mindset
19 between the -- the treating physician and the
20 examiner.

21 And if we could bridge that, we might
22 get the right answers to our questions that would

1 help us make that disability determination. I --
2 I think that's what we've been wrestling with, how
3 do we get that.

4 DR. GAHL: Well, I -- I could just
5 sort of reiterate what I said about function. In
6 other words, you're right, physicians don't think
7 about function very often. They're happy to make
8 a diagnosis and give some medications, et cetera.

9 But the -- the physical therapy and
10 the rehab folks do think about function all the
11 time. So perhaps you could require that there be
12 a rehab medicine type consultation on this, you
13 know, what can the person do and what can't the
14 person do attendant to this diagnosis.

15 What do you think about that?

16 DR. GROPMAN: Yeah. Yeah, that makes
17 sense. Yeah. I think, you know, neurologists are
18 focused somewhat on function, but certainly our
19 rehab colleagues, that's the bane of their
20 practice, and to say, okay, you know, thank you,
21 we have the information about the diagnosis, we
22 need to determine the functional limitations of

1 this individual.

2 I would think that perhaps some places
3 would not necessarily have access to physical
4 medicine, so basically laying out exactly what a
5 functional determination is might be helpful in
6 those cases.

7 COMMISSIONER ASTRUE: Do you have
8 anything else?

9 JUDGE CRISTAUDO: No.

10 COMMISSIONER ASTRUE: Do you want to
11 make any final remarks?

12 ACTING DEPUTY COMMISSIONER RUST: I --
13 I would just like to -- to thank all the witnesses
14 today, all the Panel Members, because the
15 information has been very helpful, and I think
16 it's going to -- going to go a long way to helping
17 us tackle this particular problem.

18 COMMISSIONER ASTRUE: I know, I've
19 just checked with Steve and Frank, they don't have
20 any final remarks except also to thank the
21 panelists and everybody that's participated today.

22 I just want to say it's been -- this

1 has been extraordinarily useful. Just to follow
2 up and on everything that's been said today that's
3 helpful, it's going to take a very, very long
4 time.

5 But I do want to remind people in the
6 audience that, from our vantage point, this is not
7 a one-shot exercise, and that it's our intention
8 to have these types of hearings on a quarterly
9 basis on a different -- different subject each
10 time.

11 And we have blocked out a schedule for
12 the rest of the year and have the next three
13 meetings set, and as we get into the summer, we'll
14 have the next year meetings set. And by the time
15 we've exhausted all the obvious things, there will
16 be time to go back and do updates on technologies
17 on the others.

18 So this is -- from our vantage point,
19 we're trying to fundamentally change how we access
20 medical information and -- and as you can see from
21 today, there's so much that's out there, it's so
22 hard, it's so complicated, that if we just try to

1 do it in our own closed world, as wonderful and
2 talented as our doctors are, we can't possibly do
3 it all. And so we're trying to find more ways to
4 reach out and get as much help from experts in the
5 field as possible.

6 I think this is a terrific way to
7 start. I think all four of these Panels were
8 extremely helpful to us. And so I want to thank
9 everybody who participated, Dave and Diane and
10 Glenn and the Staff that put together this
11 terrific program. And thank you all, and I -- and
12 I'll let you out 17 minutes early.

13 So, again, thank you all.

14 (Applause.)

15 (Whereupon, the Outreach Hearing
16 adjourned at 4:13 p.m.)

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Cindy L. Sebo, RMR, CSR, CRR, RPR
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