SOCIAL SECURITY ADMINISTRATION

COMPASSIONATE ALLOWANCES OUTREACH HEARING

ON RARE DISEASES

Washington, D.C.

Wednesday, December 5, 2007

The Outreach Hearing on Rare Disease

Began at 9:25 a.m.

BEFORE MEMBERS:

MICHAEL J. ASTRUE

STEPHEN GROFT

FRANK CRISTAUDO

DAVID RUST

Reported by: Kathy Savich, RPR

P R O C E E D I N G S

MS. BRAUNSTEIN: Good morning. On behalf of the Social Security Administration, I would like to welcome you. The room we are sitting in is the property of the International Trade Commission. They were gracious enough to lend us this space, and they've asked that I make the following announcement.

If you hear the sounding of an emergency alarm, please go out the back door which is straight back, then proceed to the end of the block and people will find you and give you instructions.

And there's no cell phones to be used and no food is allowed in this room. Thank you very much.

COMMISSIONER ASTRUE: Welcome to day 2 of this hearing. Before we begin, we have a slight shuffling up here. Officially, as a New Englander, I'm horrified that Dave Rust is late because of the weather.

Washington, as you know, shuts down for about two days for every inch of snow that we have. And he -- David, in his defense, is coming in from Damascus, Maryland, which is quite a distance from here. So he will be here. He's on his way. He's upset that he's not here already, but he should be coming in shortly.

We did know that Dr. Stephen Groft, who sat in yesterday and contributed so much, had a presentation this morning. They did move him from third to first, I believe, on the panel so he could get here more quickly, but we have David Eckstein from NIH sitting in in his place, so I think we'll be extremely well-served.

I recognize a lot of the people in the audience here for day 2, so I won't go through our purpose, and think we want to get to the panelists as quickly as possible and give them as much time to make their presentations and have an exchange with the panel. But I think for any of you who were

not here yesterday, our purpose here is to try to change the way in which the agency processes medical information so that we can decide disability cases more quickly and more accurately and, ultimately, that means more compassionately.

We've got several themes. We're looking for diseases and conditions that we ought to be able to fast-track. We have new computer model techniques that can cull electronically filed applications out of the system extremely efficiently, but -- using certain key terms. We're in the process of taking the first of what should be a two-track system national now. It's worked very well in New England. It's rolling out through the rest of the country.

We're looking to create a second track that should be even faster and easier for diseases where, by nature of the disease or condition, we're simply going to presume disability.

We also are looking to use biomarkers, imaging data and other new scientific developments in ways that we usually haven't used them in the past to try to draw clear lines in large diseases and conditions, to try to find subpopulations where we can say with a high degree of certainty that the subpopulations meet our statutory standard for disability.

And as an example of this that we've done recently, we've just updated our digestive regulations for the first time in a very long time, and we collapsed most forms of severe liver disease into one category and said that, using a common diagnostic scale that has three components called the MELD score, if you have a score of 22 or more, we're simply going to presume disability.

We're looking at doing more of that. We're also -- we've had great success partnering so far with NIH, and we're discussing trying to run some clinical trials

to try to tie biomarkers to functionality.

So we're looking at questions whether we can use MRI scans to identify the most seriously affected multiple sclerosis patients. We're looking at questions as to whether we can use some of the nifty new techniques in cardiac imaging to measure blood flow to the heart in perhaps the same way the we have MELD scores, say, if you're just not getting enough oxygen because you don't have enough heart function, we can't reasonably expect you to perform the functions that we use to determine whether you can engage in substantial gainful activity.

So that's essentially the purpose of today's hearing. Today is the first, but it's not a one-shot deal. This is part of a planned quarterly process. They won't all be on rare disease. The next one is on cancer. We're going to be doing psychiatric diseases, traumatic injuries, and then we'll be rolling through a regular schedule. So if you have

any interest -- the next three are set -- you should talk to Diane Braunstein, because she's coming in here, to get the details for future hearings.

So, again, I want to thank you all for coming today. I'm sure today's panelists will be as terrific as yesterday's were. I don't know -- David, Frank, do you have anything you want to say to open?

Okay. I think we're good to go, and we can welcome our panelists.

Our first panel today, we have three guests. We have Kathy Hunter who is the founder and president of the International Rett Syndrome Association. We have Craig Polhemus who is executive director of the Prader-Willi Syndrome Association, and we have Vicky Whittemore who is vice president and scientific director of the Tuberous Sclerosis Alliance.

Thank you and welcome.

Kathy, do you want to start for us

or do you have a set order, somebody who wants to start?

MS. HUNTER: I'll be happy to.

COMMISSIONER ASTRUE: Okay. Great.

MS. HUNTER: Mr. Astrue, Mr. Groft, in absence, Mr. Cristaudu, Mr. Rust and fellow --

COMMISSIONER ASTRUE: Hold on a second. We need to do a mike check, I think here. I think we have the mikes on. We discovered yesterday that you have to speak extremely close to the mikes, more so than usual so that you cannot only be heard here, but theoretically we have people listening all over the country who have dialed in. So I'll ask you to speak very close to the mikes.

MS. BRAUNSTEIN: If I could ask, there is a delicate balance. It's close, but not loud. So good luck.

COMMISSIONER ASTRUE: All right. We'll jump in if we have a problem.

MS. HUNTER: And fellow members,

thank you for this opportunity to participate in this Compassionate Allowances Outreach hearing on social security benefits for those with rare disorders. I deeply appreciate your consideration of seeking ways to better serve this population.

My comments today are based on my experience as founder and president of the International Rett Syndrome Association for the last 23 years and as the mother of a grown child, now 33 years old, with Rett syndrome.

By way of background, Rett syndrome is a genetic and neurological disorder which affects one in 15,000 females. It's seen primarily in girls who develop typically for the first 12 to 18 months of life, after which a devastating regression leads to loss of speech, mobility and hand function. Seizures, breathing problems, gastric difficulties and loss of muscle tone and ambulation are common. Rett syndrome results in severe to profound disability by the age of three years.

Individuals with Rett syndrome need assistance for every aspect of daily living for the remainder of their lives. They will never be self-supporting wage earnings.

While it is not the label, but the extent of the disability which frames eligibility with social security, in Rett syndrome, it can be assumed that any individual with a label should qualify unquestionably. However, as a rare and relatively newly discovered disorder, Rett syndrome is not widely recognized, even in the medical community at large. This makes a particular problem for families advocating for their loved ones when making application for social security benefits.

Adjudicators and even the allied medical community more often than not have never heard of Rett syndrome and may even have difficulty locating printed resources on the disorder. The typical response to saying that your child has Rett syndrome is, "What

syndrome"? This prolongs the processing of claims, not only in terms of establishing basic eligibility, but also in understanding the impact of Rett syndrome on the child and the family. Maintaining evidence of diagnosis may be further delayed because the biological marker for Rett syndrome, a mutation on the MECP2 gene of the X chromosome, is now found in only 90 percent of known cases, leaving the remaining 10 percent to a clinical diagnosis.

It would be very helpful to have a list of rare disorders in which the disability would be assumed. Hopefully, this would alleviate the huge backlog at Social Security by moving the simple cases much more expeditiously. Interviewers should be familiar with the list. Rett syndrome is often confused with Tourette syndrome, and they are two very different syndromes. And the inside joke on that we usually reply to, when people say, "Do you mean Tourette syndrome?" The response is, "No, they are

Tourette, and we are one." Rett syndrome is not what you get from watching too many reruns of Gone With The Wind. That's called humor.

The Residual Functioning Capacity Questionnaire for physicians is easily and quickly completed by hand. The problem is that they are not widely used until an appeal is in process. Interviewers should be afforded some basic sensitivity training to understand the overwhelming nature of the application process for families of children with disabilities who often have kind of been through a long ordeal before they even get to the process of applying.

Families on the income edge find their financial lives become an open book where every penny is counted, even when the monthly income stream is unstable. Some interviewers can be very intimidating. Of course, some can be very good as well.

Sensitivity, empathy, understanding and kindness are what are needed by parents

who are already stressed. Parents should not be made to feel inadequate or greedy for seeking services that will help their children. They should be given information openly, instead of reluctantly, on how to determine maximum benefits or how to play the game, so to speak, without having to go underground to learn the secret formula for increasing the amount to which they are entitled. Parents should not be punished for providing financial support for their loved ones and for keeping them at home and out of institutions.

Social security programs have an immense potential to enhance the lives of individuals with rare disorders. Your attention to streamlining and improving these programs are deeply commended. Thank you.

COMMISSIONER ASTRUE: What we're going to do as a procedure -- we saw that it worked better yesterday -- is to let each panelist speak and then hold our questions to

the end because a lot of times the questions seem to have commonality, and multiple panelists want to comment on that, so we're going to try to curb our enthusiasm until the end. Okay. Craig.

MR. POLHEMUS: First I need to say that SSI is just a vital, a tremendous, a life-saving program. It's tremendous for those populations who are able to get onto SSI. In addition to providing for Medicaid, it therefore -- it provides the funds that can be used to pay for group homes, which become necessary for most people with Prader-Willi syndrome as they reach their teen years and their adult years because their behavioral problems just become too difficult for families to -- to handle. Therefore, it's absolutely tremendous that you're undertaking this initiative.

Prader-Willi is relatively rare. It's estimated to occur in one to every 12,000 to 15,000 births. With respect to SSI, we

find that our individuals almost always lose at the initial determination stage, and we are unaware of any case, in which -- if they pursue reconsideration and appeals and hearings, we are unaware of any case in which they have lost, as we say, at the administrative judge level or earlier.

It's also very easy to establish whether someone has Prader-Willi syndrome. Genetic proof is all that is required. I know you're looking for scale in many cases -- a scale of disability. And in our case, that just doesn't work because every individual that we know of is unable to work independently, usually unable to live independently as well.

Prader-Willi syndrome is a complex genetic disorder affecting appetite, growth, metabolism, cognitive function and behavior. The most notable characteristic, the one that most people know, if they know anything at all about Prader-Willi syndrome, is that

individuals who have the syndrome never feel full. They are always hungry. One person said, "I feel that there are a thousand piranhas chewing at my stomach all the time."

What this means is that they are, therefore, unable to resist food seeking. If there is food available, they will eat. They will never stop eating.

If money is available that can be stolen to buy food, unfortunately that frequently happens as well. There is no drug to control this hunger.

The main issues in daily life for these individuals are both food-seeking and the behavioral issues. Both of these are biologically-driven, and that can be proven because individuals who have had brain injuries to the hypothalamus have -- often have what's called acquired Prader-Willi syndrome. It's not really Prader-Willi syndrome, but because the hypothalamus has been injured, the symptoms are exactly the

same, thereby demonstrating that it's not a matter of lack of self-control; it's biologically-driven.

There have been tremendous medical advances. Use of a human growth hormone as well as controlling access to food has dramatically reduced the extent of life-threatening morbid obesity among the new generation of people with Prader-Willi syndrome. But, unfortunately, that does not help in any way the food-seeking behavior or the behavioral issues. In fact, an individual who is not morbidly obese will have a smaller stomach, and if they binge, they are, therefore, more likely to have stomach ruptures, which, of course, can be fatal.

No matter how high-functioning an individual with Prader-Willi syndrome is, these behavioral flare-ups, including rage, will persist. If they're more intelligent, there will be better food-seeking. They will be more frustrated that they're unable to

behave in a normal way. And they'll be, as I say, better at stealing not just food, but also money to buy food.

This is not just a matter of while you're in a workplace; going to/coming back from a workplace is also about a place where food-seeking can occur. I suggest that if you look around your own work environments, just be -- look for food or a place where money could be stolen, I suspect you will find a surprising number of them.

Not only do the food-seeking and the behavioral issues make it basically impossible for regular employment, but many, perhaps most, sheltered workshops are also unable to accommodate the needs of people with Prader-Willi syndrome because they require constant supervision, constant making sure that no food is available. One individual who is really one of our success stories just recently was found to have been stealing food from other sheltered workshop individuals.

And the question was, "Gee, how did he possibly get unattended in order to do that?" And the answer was, "Gee, they let him go to the bathroom ten times a day." So hopefully we've convinced them that he doesn't really need to go to the bathroom ten times a day and, therefore, will no longer have that capability.

The behavioral issues include uncontrollable rage at the smallest things. They are unable to distinguish big issues from small issues. "Someone else used my broom" can be a trigger for an uncontrollable outburst. And they're also very slow at calming down from those outbursts. So that is obviously not the kind of thing that can normally be accommodated in a work setting.

As I say, your experience -- Social Security Administration's own experience demonstrates that these people are, in fact, disabled. Therefore, your disability determination processes, the appeals and the

hearings, the work of our crisis counselors who work with these people every day to encourage them to continue with their appeal processes -- all of that is wasted administrative effort since we already know the answer: If they do not give up, they are, in fact, going to be found eligible for SSI.

So, finally, I'd like to urge you not to wait. Don't let the excellent be the enemy of the good. Don't wait until you have everything in place in order to take those steps that you already can determine are appropriate to improve the determination process.

In our case, that means we're suggesting that Prader-Willi syndrome be added to the list of impairments establishing disability. There is no point in going through an individual determination of something that you know already the outcome is going to be that they are disabled -- and, as I say, using genetic proof that they have this

syndrome, this could become an easy process. Thank you.

COMMISSIONER ASTRUE: Thank you.

Vicki.

DR. WHITTEMORE: I'm Vicki Whittemore with the Tuberous Sclerosis Alliance, and Tuberous Sclerosis Complex is a genetic disorder that affects around an estimated 50,000 individuals in the United States, and it is caused by a defect in one of two genes, the TSC1 or TSC2 gene, that have been identified.

And the disease itself has really two different parts. One is caused by abnormal development of the brain that can result in an individual having seizures, learning disabilities anywhere from mild to severe, to autism, and also a significant number of psychiatric disorders such as attention deficit, depression, anxiety disorder, which are very significant in this population.

The other aspect of the disease is that these genes are known as tumor suppressor genes; in other words, when they function in a normal individual, they suppress cell growth and tumors don't form. But when there is a mutation is one of these genes, then tumors can form in multiple organ systems.

The difficult aspect about this disease is that it is highly variable from one individual with the disease to another, even within the same family. So I first heard the words Tuberous Sclerosis from my nephew who was diagnosed in 1985, and he is now 23 years old, very severely affected and has multiple organ involvement, autism, mental retardation. He functions about at the level of a two-year-old. And then, five years later, after he was born, my son -- youngest son developed seizures and was diagnosed with Tuberous Sclerosis, as was I. And so you can see within even our family how variable the disease can be.

So to address the questions that were posed to us, first, the experience of individuals with TSC, which, as I said, is a genetic disorder and affects one in 6,000 live births, or approximately 50,000 Americans, in filing for benefits is mixed. Some individuals are able to apply and receive approval for benefits as quickly as three days later, whereas others are denied and required to appeal. This is particularly true for adults with Tuberous Sclerosis who are applying for the first time as adults when their condition becomes serious or so serious that they can no longer be employed and require benefits.

Secondly, the experience of individuals with TSC and their families is that no one has ever heard of the disease and it's very often confused with tuberculosis, which is an infectious disease, not a genetic disorder.

Individuals who apply for benefits

are advised to supply as much documentation about the disease as possible with the application that describes the various and multiple manifestations of the disease, the chronic nature of these issues and the fact that these issues may be so serious as to be disabling in a majority of the individuals with TSC.

Individuals with Tuberous Sclerosis are typically followed by a neurologist because of chronic seizure disorders, and this physician is most always very knowledgeable about TSC, but most primary care physicians are not so familiar with the disease.

As I say, adjudicators are not familiar with TS, and the individuals applying must supply documentation of the disease, extensive medical documentation regarding their disabilities that are caused by the disease.

Third, the -- TSC may be a terminal illness for some individuals, but the TSC

community does not have experience with this aspect of processing claims since it's most often considered a chronic disorder, not a terminal illness. TSC can be terminal, however, because of the multisystem nature of the disease affecting the brain, heart, eyes, kidney, lungs, liver, skin -- virtually any organ of the body. The disease can be life-threatening if appropriate diagnosis and treatment of the symptoms of the disease is not received.

Since there are no treatments specifically for TS, individuals with the disease are treated for each of the symptoms of the disease: Seizures, autism, brain, kidney, heart, skin, liver tumors, learning disabilities ranging from minor to severe and, as I said, mental health issues, including depression, bipolar disorder, anxiety disorder, obsessive-compulsive disorder and attention deficit hyperactivity disorder.

To answer the fourth question, the

current system would be greatly helped by access to a list of rare disorders that describes the potential impact of the disease, the variable manifestations of the disease and their presentation over the lifespan of individuals with the disease, and also the details, the objective medical evidence needed to establish the condition.

This would alleviate the need for individuals with the disease to acquire and provide so much information about the disease itself to SSA adjudicators, and would help them by specifically identifying the information needed for their claim. The problem with many rare diseases, as I've already pointed out, including TS, is that they can be highly variable from one individual to the next, even within the same family. This is often a problem for adjudicators to understand and to obtain accurate information about the variable nature of the disease.

An individual with TS may be mildly affected as a child and not require benefits. But some of the manifestations of the disease become significant during adolescence and adulthood. Having information to this effect would be very helpful for adjudicators.

The diagnosis of many rare disorders has evolved significantly over the last 10 to 15 years such that now individuals who are mildly affected by the disease are being identified and diagnosed with a particular rare disease whereas, in the past, only the most severely affected and disabled individuals were diagnosed, and this is significantly the case with Tuberous Sclerosis.

The mild affected individuals may have significant disability from one aspect of the disease even though they are unaffected by other aspects of Tuberous Sclerosis.

In addition, since research is moving so rapidly for many of the rare

diseases with the identification of the gene, or genes causing the disease, elucidation of the function of these genes and the development of treatments leading to clinical trials, the landscape for treatment will also be changing significantly. Providing an avenue for updating and providing new information to the SSA from the respective rare disease research and clinical communities in a clear and concise manner would be beneficial.

Also, most of the rare disease nonprofit organizations, like the Tuberous Sclerosis Alliance, have volunteer professional advisory boards composed of knowledgeable healthcare professionals who can serve as a resource to the SSA as needed to assist in obtaining information about a specific rare disease.

And, last, the Tuberous Sclerosis Alliance greatly appreciates the opportunity to participate in this hearing and applauds

the SSA on this initiative to approve service for individuals with rare diseases. These individuals and their families often struggle just to get a diagnosis of their disease, and having a quick and accurate review of their SSA claims would be beneficial.

Individuals with rare diseases and their families are very often juggling medical, educational, financial and employment issues. Clearly defining the medical evidence needed, providing lists and information about rare diseases to the adjudicators and providing a resource through the nonprofit organizations to healthcare professionals who can assist the adjudicators when needed would greatly improve the system.

In addition, providing a mechanism by which new information can be provided to the SSA as new diagnostic methods are developed, new information about the disease and new treatments come online would also significantly benefit the processing of

claims. Thank you very much.

COMMISSIONER ASTRUE: Thank you very much.

Before I launch into a few questions, let me say a couple of things. First of all, I wanted to respond to Craig's point that -- certainly share the sense of urgency. So for people in the audience who were not listening yesterday, our general time frame for creating the -- we've got the QDD process, which will be the slightly slower and less automatic of the two fast tracks. That's up and running now. The last report I had we were at 28 states up and running. That should be pretty much nationwide within a matter of a few weeks.

The Compassionate Allowance track -- probably a reasonable expectation would be sometime around Labor Day of next year. It might be a little bit sooner. It is important, when we're doing new things, to do them right. We have a little bit of a track

record of the best of intentions, rushing things, and then creating problems in the field. At the end of the day, we don't do anyone any favors if we don't have our systems tested properly, we don't have the training done and that type of thing.

It may be possible to give some guidance to the field before that so that there's something transitional, perhaps in a commissioner's ruling -- and we're talking about that, and it may be possible to do that relatively promptly. So we are looking at the possibility of that as well, but it shouldn't be extremely long timelines.

And the other thing I just want to say is we are focused here primarily on trying to come up with a Compassionate Allowance list, but we realize that there are a number of very serious diseases where, because there is a moderate population, often a very small part, we probably have it on the QDD list rather than Compassionate Allowance.

And there will also be a few diseases that we're looking at in this panel where, because it is complex and because there is a spectrum, we may only be able to put a subpopulation on one of those lists, and it may be that the ultimate answer is we just have to give a lot more detail in our listings the way we do for diseases generally. We have very few rare diseases in our listings in general.

So there are basically three buckets, and although we've advertised this as a Compassionate Allowance hearing, we're not going to be fussy about that. I think we're just trying to figure out, you know, the right thing to do for as many of the rare disease populations as possible. So that may be helpful in terms of clarifying where we are and what we're trying to do.

Let me start with the disease I'm by far the most familiar with, which is Rett syndrome.

MS. HUNTER: What a blessing to hear you say that.

COMMISSIONER ASTRUE: Well, when I was a biotech CEO, your association -- some parents came in and spent a morning with me and my scientists, and we took a very serious look at it. And it was very frustrating because we concluded, (A), that we could make the protein properly and then, (B), we were persuaded -- we were outliers on that, but we could administer protein to the brain. We were looking at modifying the Oliver shunts [phonetic] that they use for oncology to administer protein in the brain. And I believe the company that bought my company is still on the track to do that fairly soon.

I think the technical problem we ran up against is it turns out that, unlike most of the protein deficiencies, you just need huge quantities of protein. And our technical people came to the conclusion that we just couldn't, through that mechanism,

administer that much protein in the brain. So it was very frustrating because we got some initial threshold answers that were positive, and then we got basically to the last technical issue and just decided it was a no-go.

But we did spend several weeks in analysis on that, so it's one that I'm a little bit more familiar with.

And I think this is one of the -- ought to be one of the easier cases -- in fact, we've been using it as an example. And I know that we've got the National Association of Disability Examiners in the back of the room there -- and, actually, when I was speaking to them in South Dakota three or four months ago, I used this as one of the examples. I actually asked the audience, how many of the examiners had actually seen a Rett syndrome case over the course of their careers, and about half of them raised their hand and said that they had seen it, but half

of them had never seen a Rett syndrome case.

MS. HUNTER: Wow.

COMMISSIONER ASTRUE: I guess the big question I have for you -- I mean, the course of the disease is pretty clear in most of these cases, and for most of the patients, the diagnosis is pretty clear. You know, from our parochial point of view, that just makes life easier, for the most part, for us. The only thing that I really see, off the top of my head, as a complication is you mentioned that 10 percent of the patients where it's not a genetic diagnosis.

If you could talk to me a little bit more about how those patients are diagnosed, if there's any science underlying, you know, that, and whether the course of the disease is any different for that 10 percent of the population.

MS. HUNTER: First of all, as the mother of the first child in the United States diagnosed with Rett syndrome, there are no

more wonderful words to hear than, "I'm most familiar with Rett syndrome." That's just amazing to me, and I am most happy in a room full of people talking about and educating them about Rett syndrome.

COMMISSIONER ASTRUE: It's a credit to your group because you came and found me, you know, three, four years ago. So it's nothing I did. It was all -- you know, you were doing the right things in trying to reach out to people that might be doing research in the area, so it's really --

MS. HUNTER: Yes.

COMMISSIONER ASTRUE: -- all a credit to your colleagues.

MS. HUNTER: Yes. Thank you.

Right now, it's between 90 and 95 percent of girls who actually get the molecular diagnosis, and the remaining 5 percent probably have just a more obscure mutation. There are more than 200 different mutations on the MECP2 gene. And these are

found in eight different hot spots.

Some children will actually have their own solitary mutation that is independent, and so that causes more difficulty in terms of research when they have a spontaneous mutation; that is, when you have a rare disorder, and then a rare mutation within that disorder.

But the treatment is no different; it's just that the MECP2 gene has also been connected to late orders of -- disorders of late development, such as schizophrenia, autism, bipolar disorder. So the MECP2 mutation can result in Rett syndrome. It can also result in other disorders. So in order to get the diagnosis of Rett syndrome, you must have the mutation, and you must meet the clinical criteria.

However, you can meet the clinical criteria and not have the mutation because we have a 95 percent rate of finding mutations. And the other -- that other subgroup of

children, 5 percent, you know, they fulfill a diagnostic criteria, and years ago, that was all we had to make the diagnosis of Rett syndrome. So it's just a matter of refining technology to be able to find the mutations in those remaining 5 percent.

COMMISSIONER ASTRUE: For that 5 percent, is the natural history of the disease pretty much the same or is --

MS. HUNTER: Yes. Oh, yes.

COMMISSIONER ASTRUE: It's not --

MS. HUNTER: Yes.

COMMISSIONER ASTRUE: I mean, it's fairly common in protein deficiencies --

MS. HUNTER: Yes.

COMMISSIONER ASTRUE: -- that you have at least, in some cases, a more moderate group where there is some residual capacity to make protein. But the --

MS. HUNTER: Well, we definitely find there are some mutations that are related to more severe cases. We have a

genotype/phenotype database that is -- so we do find that there are some mutations that present as milder.

The complicating factor is that the severity really is determined by the activation rate, which is independent of what the mutation is, so there are two factors to consider. So you can have two children with the same mutation who present very differently.

But, you know, in terms of qualifying for SSI, the majority of these children are not nonverbal, completely nonverbal. A few of them have single words, a few phrases, but even those who do have a few words, "mommy," "food," you know, very basic things, you know, they're not functional with language.

And about half of them are able to walk. None of them have functional use of their hands, so they're not able to work without hand-over-hand assistance. So it's

really -- probably should be a very clear one that someone -- a clear case that someone with Rett syndrome should qualify.

COMMISSIONER ASTRUE: Let me move on to Craig's. I'm less familiar with Prader-Willi. If you could talk to me a little bit more -- we have to make some -- we have a very demanding statutory statute here. The 12 months, you know, to perform work and the economy -- and you mentioned people in the workplace stealing and -- stealing food and that type of thing.

Can you talk to me a little bit more about what percentage of the population is in the workplace, what types of job functions they tend to perform and how long they tend to last in the work force and that type of thing.

MR. POLHEMUS: If you're talking about regular employment, not sheltered employment --

COMMISSIONER ASTRUE: Yeah.

MR. POLHEMUS: -- there's very, very little. You know, you've made it through school because you can get even a one-on-one aide, if necessary, but that's basically what you would need in the workplace as well.

So -- I mean, we have people who can work for churches. Often it's volunteer; occasionally they get paid for it. We did have someone who was stocking in Target, but lo and behold, when he was stocking cookies, it ended up he was eating the cookies. You know, if anyone leaves their locker unattended, it's going to be opened up and they'll look for food or they'll look for money in order to buy food.

So if you -- we have some parents who take their kids to the workplace -- and, in fact, this applies to school as well -- take their kids to the workplace every day, pick them up, bring them back every day. But unless they stay there with them, or have someone else stay there with them, the

likelihood that they'll continue in employment -- I'm not aware of anyone who has lasted a full year.

COMMISSIONER ASTRUE: So that's pretty much -- and there isn't a milder population that has some success in, you know, limited employment now? I mean --

MR. POLHEMUS: No, there is not. And, in fact, in most cases they're also unable to -- I mean, obviously you can't live independently either. You need 24-hour supervision. The group home has to lock their kitchens and lock their cabinets.

COMMISSIONER ASTRUE: Just one other question I wanted to ask, just because I was curious. Is this a protein deficiency or do they know the mechanism of action for the disease?

MR. POLHEMUS: No. It's a defect on chromosome 15. It's actually the internal chromosome 15 which doesn't get activated. There are at least three genes that are

implicated, and the main impact is on the hypothalamus.

Now, I focused on food-seeking and behavioral flare-ups because those are the ones that I felt were more significant to work. But, in fact, there is a wide range: Apnea, apraxia, scoliosis, dental and saliva. Almost every -- reaction to anesthetics. In almost every field of health, of medical, they have special challenges.

So I don't believe that it's anything as easy as a protein deficiency.

COMMISSIONER ASTRUE: Okay. That's helpful. Thank you.

Vicky, just a couple of questions for you. You made it clear from your presentation that there is much more of a range of functionality. If you could give me a little bit more of a sense of -- let's take the adult population first, just to try to make our exercise a little easier.

Do you have any sense of what --

you know, right now, what percentage of the adult population is performing work outside of a sheltered workshop, is performing regular jobs in the national economy?

DR. WHITTEMORE: I would say about 50 percent --

COMMISSIONER ASTRUE: About 50?

DR. WHITTEMORE: Yes, about 50 percent can live and function independently. Some of those individuals, however, will develop kidney tumors down the line and become disabled but, you know, they're able to work up until that point until they -- because they have very little brain involvement. So -- but I would say probably about 50 percent.

COMMISSIONER ASTRUE: So let me ask you, again, for diseases and conditions like this, almost inevitably there are going to be some judgment calls and some close calls, and we can't eliminate that from the system entirely.

DR. WHITTEMORE: Sure.

COMMISSIONER ASTRUE: But to the extent we can draw some lines, you know, to make some of the cases automatic, that would certainly, you know, be helpful.

Is there any biomarker or any MRI test or anything that we could look at that you know of that would give us a sense for the people that are most severe, that, you know, would give us a basis for saying, "yes, this is somebody that would be unable to work"?

DR. WHITTEMORE: Well, there is a genetic test, but that doesn't tell you anything. The two genes are quite large, and we now have over 2,000 mutations identified in those two genes. So most families or individuals have their own private mutation.

COMMISSIONER ASTRUE: Right.

DR. WHITTEMORE: So that doesn't help you much. And mutations in one gene versus the other also doesn't help. There is some slight indication that if you have a mutation in the TSC2 gene, you will have a

more severe form of the disease, but that does not carry weight.

In terms of MRI, overall, there is a correlation between brain lesions, that are called tubers -- they are sort of like birthmarks in the brain; they're actually malformed areas of the cortex that are there at birth. And so the more of those tubers or brain lesions you have, the more likely you are to have early-onset seizures and then lifelong learning disabilities anywhere from mild to severe mental disabilities.

So that is a fairly good biomarker, or marker, would be to go based on the brain MRI. But it's not 100 percent.

I know a girl who is graduating from high school who, if you looked at her MRI, you would be astounded that she is even able to walk and talk. So it's pretty clear-cut, but not 100 percent.

COMMISSIONER ASTRUE: Let me ask a variant on the same question. So you

mentioned -- and I'm not sure I got it all; I jotted a short list here. The range of other -- seizure, brain, kidney issues, that kind of thing -- that are associated with more severe forms of the disease.

For the patients that seem to be heading, for instance, toward a terminal condition, is there any pattern in terms of they've got the underlying disease, and they develop the kidney issues or they develop something else in particular -- as an example of what I'm thinking about, for long time I worked on Fabry Disease, and, you know, it has a lot of horrible symptoms, but a lot of the patients live fairly long, even before there was enzyme replacement therapy.

And the three main causes of death -- stroke was unpredictable. Heart was unpredictable. Even though they would get swelling of the heart, you didn't seem to be able to correlate -- it seemed like it ought to correlate. You would have a very large

heart, but you couldn't predict a heart attack.

But on the kidney, you could say with a fair amount of precision, once they lost a certain amount of kidney function, they were heading toward dialysis and death within a fairly predictable period of time.

Is there anything like that for the most severe patients that we could be looking at?

DR. WHITTEMORE: Yes. Most of them will succumb to either a brain or a kidney tumor. So generally the kidneys become overwhelmed by these very large benign tumors, and then that leads then to individuals requiring dialysis and, eventually end-stage renal failure. So, yeah, those two things probably.

COMMISSIONER ASTRUE: Okay. That's helpful. All right. I am hogging your time. David, do you have questions you want to ask? None right now?

Frank.

JUDGE CRISTAUDO: Yes, thank you very much. I appreciate the comments. I just want to clarify a couple of points and add just some further questions.

For the impairments that Ms. Hunter and Mr. Polhemus talked to us about, it's pretty clear, in their impression, that everyone who has a diagnosis is essentially disabled. Did I understand that correctly?

MS. HUNTER: Very clearly.

MR. POLHEMUS: Yes.

JUDGE CRISTAUDO: Okay. And certainly not, as Dr. Whittemore -- it's not the same.

Mr. Polhemus did mention to us that a number of the cases actually get to the hearing level in our process, but once they get to the hearing level, they're always approved, I think is what you said.

MR. POLHEMUS: I'm unaware of any case that was not approved, that's correct.

JUDGE CRISTAUDO: Okay. And, Ms. Hunter, with --

MS. HUNTER: The same. They're usually approved.

JUDGE CRISTAUDO: But some need to get to the hearing level at this point?

MS. HUNTER: That has happened. In the majority of the cases, it's fairly clear, but some have reached the hearing.

JUDGE CRISTAUDO: But your sense is that most do not actually --

MS. HUNTER: Most do not.

JUDGE CRISTAUDO: -- get to the hearing level; they're decided before.

And then, Dr. Whittemore, the cases that get to the -- well, actually, the question is for all of you. The cases that actually get to the hearing level, it's pretty clear that, in the first two situations, they're generally approved. With the impairment that you're representing, what's the experience that you're aware of?

DR. WHITTEMORE: The experience is that it's, again, mixed. Many individuals will have been functioning normally, working, and then have -- you know, end up with chronic kidney problems, needing dialysis, having a severe disability, for example, and/or severe disability from seizure disorder that they now cannot control and can't work.

And those are very often denied at the hearing because a person has worked in the past and has been able to live independently in the past.

So, you know, sometimes those are approved and sometimes not. It's very mixed.

JUDGE CRISTAUDO: Okay. And the commissioner has made it very clear that certainly the agency is looking at if there's some way that we can identify additional impairments that we can look at and simply approve because we know that, in fact, people will become disabled at some point. But we still are going on, at this point, certainly

with cases to the hearing level.

Is there something that we should be asking about, if the case reaches the hearing level, so that we can get some additional information at that level even at this point -- that we could get that information and make a decision very quickly without even having to go to a hearing? And that's really for all three of you. If you can think of any information that we could be asking for or anything we could be doing to help us establish pretty quickly, just looking at the record without having a whole hearing.

MR. POLHEMUS: In our context, I'm not really sure what that question means. It would seem to me that genetic proof of Prader-Willi Syndrome, based on your own experience, would be sufficient. I don't know that there are any additional questions that you would need to ask.

JUDGE CRISTAUDO: Okay. And then probably the final question, but kind of a

related question: Is there additional information we should be requesting, additional forms we should be asking people to fill out? Should we be doing anything else at the early stage of the process to make sure that the adjudicators are very familiar with the extent of the issue in the individual case?

COMMISSIONER ASTRUE: Can I also make a friendly amendment to Judge Cristaudo's question? I assume, for Rett syndrome, that it's always going to be the case that it's a family member filling out the application, or some other representative. If you have any sense -- particularly, I guess, for the Prader-Willi patients. Do the patients try to fill out the applications themselves or is it usually family members that do that? Or somebody --

MR. POLHEMUS: It's the parents, and often with the assistance of our crisis counselors. What our crisis counselors have

to do, generally, is convince the parents not to give up just because they were turned down at one level.

But, no, I don't think anyone would try to fill out the forms themselves.

COMMISSIONER ASTRUE: So back to the question, is there anything -- Frank's question: Is there anything else in terms of documentation or solicitation from -- that we should be asking for more systematically that, from your vantage point, would help the process? Anything that you can see in terms of --

MR. POLHEMUS: It's certainly very hard for me to do that because it sounds like you're trying to distinguish between those people with a particular condition who are disabled and those people with the same condition who are not disabled, and that's not a distinction that seems to have any relevance in our case.

COMMISSIONER ASTRUE: How about --

DR. WHITTEMORE: Also, it doesn't in our case. And I think -- you know, we encourage individuals to provide as much information that we provide to them about, you know, the chronic nature of the disease. So once you have an onset of kidney -- severe kidney disease, it's not like it's going to get better.

And so I think asking for that documentation up front about the natural course of -- natural history of the disease would be helpful. We -- the individuals who contact us ahead of time we help provide that information.

I think the people who are denied or have to appeal or go to hearing are those individuals who walk in thinking -- you know, sort of do the application themselves, walk in thinking that this is a slam dunk, and not understanding that they need to provide that kind of documentation at the earliest stages.

COMMISSIONER ASTRUE: As I have

been mulling this, I think one of the things that you could do for us that would be most helpful is -- you know, I'm sure you've got physicians that are working closely -- working closely with you and trying to be advocates from the population. If there's anything in the medical journals, studies, anything that shows what the natural history looks like -- people that have the underlying condition, and then also present the kidney tumors and/or the brain tumors, you know, that might be the kind of thing that would allow us to at least issue some bright-line guidelines for at least a subpopulation of the overall population.

Again, it's not a total answer, but at least it might --

DR. WHITTEMORE: Sure.

COMMISSIONER ASTRUE: -- help a certain segment of the population.

DR. WHITTEMORE: Absolutely.

COMMISSIONER ASTRUE: Great.

David, do you need anything else or

are you still okay? You're still fine? Frank, do you need anything else?

Okay. We're running a little ahead of schedule because we started early. Thank you very much. As with yesterday's panels, this is very -- both interesting and helpful. We're very grateful. We appreciate it.

And we'll take a 15-minute break, and we'll reconvene at 10:30 for the next panel.

(Recess.)

COMMISSIONER ASTRUE: I think we're going to try to start the next panel.

I understand there have been some problems hearing in the back, so -- the continuing technology issues, so we're going to try to -- for the panel, we're going to try a mobile microphone. We're going to turn off the microphones we have been using and try to use this instead when we're speaking, which would probably also keep us from talking over each other, too, which is probably mostly my

fault.

Okay. For the second panel, we have Suzanne Pattee, vice president of regulatory and patient affairs, Cystic Fibrosis Foundation. Dr. Michael Boyle, director of the Johns Hopkins Adult Cystic Fibrosis Program. Barbara Boyle, national executive director and CEO, Huntington's Disease Society of America. And Pat Furlong, president of the Parent Project for Muscular Dystrophy.

And before you actually start, let me do another technology check.

Can you hear me better in the back now? That's good? Okay. Let's roll.

MS. PATTEE: Thank you, Commissioner Astrue. I'm Suzanne Pattee with the Cystic Fibrosis Foundation. We're very pleased to be here today. This is a very important issue for our -- people in the community with cystic fibrosis.

I would like to turn it over now to

Dr. Michael Boyle, but before I do so, I wanted to mention that I have cystic fibrosis as well as diabetes, which is related to cystic fibrosis. I am fortunate to be able to work full-time, but only because of a lot of the medical care that's gone on prior to that.

DR. BOYLE: Thank you very much for the invitation today. My background is I run the Adult Cystic Fibrosis Clinic at Johns Hopkins, which is one of the largest adult CF program in the country. We care for about 200 adults with CF.

I just want to start out by saying thank you for being such an important part of our program even though you're here in D.C., certainly it makes possible support for a good number of our patients. You know, if you look nationally at the cystic fibrosis registry and national database, about 40 to 50 percent of individuals with CF have benefits.

And just backing up a little bit, quick reminder about what cystic fibrosis is,

although you're probably fairly familiar with it already. Cystic fibrosis affects about 30,000 individuals in the United States. It's probably the most common lethal autosomal recessive disorder in Caucasians in the United States that's caused by group of mutations -- it's not a single mutation; actually, about 1500 different mutations which can lead to dysfunction of the cystic fibrosis protein, that protein which lines most of the lumens of our body and controls a lot of the salt and water balance there.

And I think one of the key things to remember about cystic fibrosis is it's a multisystem disorder. And it's very easy to focus in on the lung part, because obviously that's the part that leads to a lot of morbidity and mortality, but definitely a multisystem disorder. So not only do you have obstructive lung disease, but chronic lung infections and chronic sinusitis, even -- as opposed to other lung diseases where there are

periods where you're well, cystic fibrosis, you never clear that infection. It's always present in the lungs and sinuses.

And those are -- those chronic times are interspersed with exacerbations which basically are pneumonia-like with increased pulmonary symptoms, shortness of breath, increased cough and sputum.

Along with the pulmonary problems, there's also pancreatic issues. So about 85 to 90 percent of individuals with cystic fibrosis have pancreatic insufficiency, meaning they need to take enzymes in order to digest their food. Even with taking those enzymes, they have a real problem with maintaining their weight, keeping their nutritional status adequate.

And then as they get later in life -- and when I say "later in life," for CF that means over the age of 18 -- they often have complications including diabetes, insulin-dependent diabetes, osteoporosis, as

well as other complications such as cirrhosis in about 1 to 2 percent of patients.

The good news is, as you've probably heard, there is increased survival in cystic fibrosis -- if you look back or talk to families when they had kids that were diagnosed with CF in the '60s or late '50s -- if it was in the '50s, they were often told they wouldn't live to see their tenth birthday. In the '60s they were told they would be lucky if they made it into their teens.

The exciting part now is that because of a lot of treatment improvement, we're up to a median predicted survival of about 36 years of age. But, you know, it's a little bit misleading because our median population age is about 16 years of age, so it's still a pretty young group.

The good news is with some of the improvements in survival, it's becoming not just a pediatric disease anymore, that over

the next decade, more than 50 percent of all individuals with CF will be adults. Like I say, I take care of nothing but adults. So that's the good news.

They bad news is that the way we're getting there is there's a real burden of treatments. So a lot of our patients spend two to three hours a day doing medications to try to, you know, have some quality of life, Suzanne can speak to this. That -- what that typically looks like is a morning where they start off doing inhaled mucolytics, spending 10 or 15 minutes inhaling a medicine to break up mucous, followed by another 20 minutes to a half an hour using airway clearance techniques, either a vest or there's other techniques that are available to try to clear up mucous, followed by another 20 minutes to 30 minutes of inhaling antibiotics.

Throughout the day, they're taking enzymes with each meal, trying to eat five or six times a day to try to maintain their

weight, and then repeat that whole process at night. As a matter of fact, I was talking to Suzanne -- I had written an article recently that was entitled, "So many drugs, so little time," which was the future of adult CF care, which is -- the great news: You know, our patients are living longer, but there's a real burden of care.

And the challenging part is even when doing that -- it's sort of, on average, maybe two times a year they end up needing to do courses of antibiotics for two to three weeks because of these exacerbations. That certainly leads to difficulties with working, being able to, you know, function normally.

And I guess I didn't really mention a lot of the energy -- the problems that are leading up to the time that they're sick and around the time they're doing medications.

So that was a maybe not so short recap of all the things that are going on in terms of the medical part of CF. But I think

one of things, just from my experience, that I wanted to stress today is it's easy to focus on the FEV1 part because we think of cystic fibrosis as a lung disease, and we think, "oh, I know that," and it's an obstructive lung disease. I look at the FEV1 and can tell what's going on. And that couldn't be further from the truth.

It's easy when the FEV1 is low, but the truth is there's lot going on before then, and, unfortunately, some of our patients have been in a situation where they're having to make a choice between trying to work or actually taking care of themselves. So I think we just want to make sure -- and Suzanne is going to talk about this somewhat -- that we address not only the end-stage, "yes, your FEV1 is horrible; you need a lung transplant," but also those years right before that when there's a real burden of care that makes it hard for them to work.

MS. PATTEE: Thanks, Dr. Boyle. So

we wanted to touch on the listing specifically. We focused primarily on the respiratory listings. We did participate recently in the review process and the comment period to advise people on the respiratory listings.

The first listing, part A, addresses the FEV1. And we estimate that someone would have to be about a 45 percent of lung function level for their FEV1 to be clearly disabled, according to the listing. But there's a separate part B and a part C. Part B asks how many physician interventions you have had in the last year. I think you have to have two -- more than two in the last six months. We wanted to talk about physician intervention as opposed to a hospitalization since technology is changing and not everyone is hospitalized, and so you can get home intravenous care as well.

And then, as Dr. Boyle mentioned, we have, under C, use an inhaled antimicrobial

medicine. There's -- antimicrobial is another way of doing an antibiotic, but because it's a lung disease, it's inhaled into the lungs directly. And people with CF, as they get more progressively sicker, often use an inhaled antimicrobial product to fight the chronic lung infection in their lungs.

So just because -- you have to meet one of the three criteria in order to be considered stable, and we're finding that many people do meet one of these three criteria but are often being denied or being told they need to wait for a hearing.

DR. BOYLE: I was going to highlight just a common experience of some of our patients. What we'll usually say to them is, you know, if your FEV1 is over 30 percent of predicted, you will be -- it will be a pretty quick turnaround if you apply for disability.

Frequently, if your FEV1 doesn't meet that obvious criteria, we just tell them,

"you know what, you're going to have to wait." You realize they're not going to get approval despite the fact that, when you look through their medical records, they really can't work. They are spending a couple of times a year in the hospital or at home, doing home IV antibiotics. They're doing a couple of hours a day of treatment to try to maintain their health.

And we're telling them, "you need to do more of this because you're slipping." That -- one case in particular that comes to mind is a 40-year-old woman who actually worked full-time for most of her life, but obviously was starting to decline, hadn't quite met the FEV1 criteria, but ended up spending two years waiting and really struggling to try to make ends meet and cut corners on her own care because she was in that period where, yes, she had been doing lots of home IV antibiotics, obviously very sick, obviously slipping and spending a lot of

time with that, but not meeting the FEV1 criteria and, so, having to wait.

MS. PATTEE: I think the main portion of this is that we have had cases, primarily if someone meets the FEV1, they are --

(Discussion off the record.)

MS. PATTEE: Often people who do meet the FEV1 part of the listings will be approved. But we do have -- know of two cases currently pending where the individuals have been waiting now almost two years for hearings, and they do meet -- as far as the listings. And oftentimes when someone will get to their hearing -- oftentimes when they will get to the judge, the judge will look at the record and say, "this person meets the listings, and so it's obvious; let's approve it." And that's disheartening to the individual who has had to wait two years for that decision.

So it doesn't always work that

easily, and then it's more difficult if they don't meet the FEV1 but do meet the two where someone might say, "well, you still don't meet FEV1," but we're saying, "that's not -- it's not an additional test. It's separate -- three separate tests."

So to the extent that the listings could clarify that these three different tests are separate and they're not additive, as well as trying to get a better picture that somebody who has a chronic lung infection who meets perhaps taking inhaled antimicrobial medicine, that is an indication of their level of involvement of their disease and that they need to be considered and evaluated as disabled because they do meet the listings.

So we have some recommendations for the system, to change it. We think it would be helpful to include some sort of lay language that would provide some of this narrative that we're providing you with to explain what the disease is -- and, yes, it is

variable, but because we have a really good education system in process with our care centers, we believe that over 90 percent of the patients who do apply for disability benefits are going to be found eligible immediately on the listings. That's not how it's being done, unfortunately.

We have developed some materials to educate the physicians and provide letters for the physicians to send to Social Security to actually document how their listings are met in the medical records. And then, as Dr. Boyle has said, he can tell someone to apply or not to apply based on his knowledge of the listing as well. So that's why we think it's a really high rate of applicants to apply.

We also would hope that anybody who -- at the Social Security Administration who may be considering denying an applicant, that they will consider consulting a physician, an expert with cystic fibrosis

because, as you know today, it's rare diseases. There are only 30,000 patients in the country. We estimate an average of 700 applicants a year who have CF. So it's rare that an ALJ would see more than one case in his entire career.

We would also like to emphasize that it's a multiorgan disease. We've been looking at the digestive system listings. Right now, we think they're pretty severe, and recommended in a Compassionate Allowance letter that if you meet a certain eligible level of body mass index -- that we think it's too strict in the digestive listings -- that it be considered to be a little more lenient.

I think right now the digestive listing is just for children's, not for adults. So you have to meet a 3 percent BMI or so, and that is almost in someone -- we rarely see people these days who meet that because we're really trying to keep health on patients. So we don't want somebody in that

state because they can't recover, basically, and they're really not going to be able to recover from that.

So we would hope that Social Security would consider assigning experts to rare diseases -- to CF specifically -- so that people who have the knowledge at the Administration can really look at these cases and be able to evaluate them more specifically and make sure they do meet the listings.

So that's our main application. Did you want to stress anything?

DR. BOYLE: We have a couple other suggestions later on for specific things. I was getting the question, from listening in the earlier time, about other things you can ask during those hearings. Do you want to hear about those now?

COMMISSIONER ASTRUE: Sure.

DR. BOYLE: So some of those things -- you know, certainly, hospitalization is an easy one, and that's already asked

about. But, truthfully, the use of home IV antibiotics for weeks at a time is something that's becoming increasingly frequent, something that's driven by insurance issues. And certainly that would be something that would help get a feel for how much time those people are having to spend on their care and how sick they are. And that's something I don't think is currently always captured.

The frequency that they're taking inhalant antibiotics to maintain BMI, they are all signs of severity of illness and are other things that you may want to be collecting at the time of getting information.

MS. PATTEE: The adult respiratory listings don't really get into some of the digestive problems. It's much more clear in the listings for children's, so we would like to see that more clearly referenced as well.

DR. BOYLE: And I think some of the education, actually, for the physician side as well -- I have to say, we have numerous

patients who are referred to us who obviously would benefit from being on disability, but have been told not until it's time to -- not until you're ready for a lung transplant, until your lung disease is end-stage. So that's, I think, one of the main things, is trying to increase that understanding not only for people hearing the cases, but for us to be able to know who to refer.

And then I guess the last part is my understanding right now is the listings seem to be about heart-lung transplantation, and just on the medical side, we rarely do heart-lung transplantation anymore just because it uses up more organs that we need to, and for cystic fibrosis we almost always do lung transplantation alone, so changing that listing would be important.

MS. PATTEE: I think that's all. Thank you.

COMMISSIONER ASTRUE: Thank you very much.

Barbara.

MS. BOYLE: My name is Barbara Boyle. I'm the CEO of the Huntington's Disease Society, and I am here with Dr. Andrew Feigin, who is director of our center at North Shore Hospital, and he is the professor of neurology at New York University School of Medicine. We would like to personally thank you for the opportunity to speak on this issue. And we are -- we greatly appreciate you looking at better ways to serve the rare disease community, which we believe is very, very important.

I want to give you a little background on what Huntington's Disease is. Huntington's Disease is a fatal, hereditary brain disease that slowly robs the affected individual of his ability to walk, talk, reason and act socially.

There are over 30,000 Americans today that have HD and another 200,000 that are at risk for inheriting this disease. HD

strikes in mid-life between the ages of 30 and 50, when an individual should be most productive. And it interferes with this individual's ability to care for his family and to work to provide for that family.

The disease takes from 10 to 20 years to progress to the end stage, at which time a person succumbs to the complications associated with HD.

HD is caused by a single defect in a single gene on a single chromosome. Those who inherit this defective form of the gene will at some point develop Huntington's Disease, and they can pass it on to each of their children.

Though the gene that causes HD was found in 1993, there still remains no effective treatment and no cure. Treatment is symptomatic and very limited. And because HD strikes so early in life, many medical and social services are not available for the people with HD and their families.

HDSA, that I represent, was founded in 1967, shortly after Woody Guthrie lost his battle with Huntington's Disease. His wife, Marjorie, gathered families from around the United States, and during the years -- 16 years that she lived -- until her death in 1993, she worked to obtain funding for HD and, frankly, family services that were nonexistent at that time.

Through her efforts, we have become the only HD organization in the United States that's dedicated to both the care and cure of our families with Huntington's Disease. HDSA funds an array of family services and educational programs. We provide access to social workers, community-based resources and referrals.

We have 21 designated medical facilities that have expertise in Huntington's Disease, and the social service workers at our centers frequently work with the families to complete the SSDI application, and our

neurologists frequently are called upon to complete the medical evaluation. But there are only 21, and we have patients all across the United States.

Our experience under ordinary circumstances is that our families report they can wait up to a year or more before their application is reviewed. And many are individuals. Family, social workers and medical professionals have recorded a variety of difficulties in receiving a determination of eligibility for SSDI benefits due to the general unfamiliarity with HD on the part of SSA, educators or physicians assigned to review the case.

At present, there exists within the system a lack of sufficient accurate medical information for evaluation of Huntington's Disease. Though the cognitive and psychiatric symptoms usually precede physical symptoms, over 50 percent of our patients have psychiatric and cognitive symptoms before the

physical chorea even occurs, and these are very disabling.

The determination by the SSA examiners and physicians assigned to these cases relies substantially upon the presence of this movement disorder. The belief on the part of social security caseworker that the applicant or his or her symptoms will improve with treatment exists. The patient's lack of insight, which is a symptom of the disease itself, about the progression of the disease results that the affected individual is incapable of giving an accurate assessment of his or her condition to the caseworker.

And the biggest common practice of our patients is that they -- they downplay reality and they downplay the extent of their condition.

Difficulty in pinpointing exactly when a person with a neurodegenerative disease first becomes disabled exists. And I can supply countless of examples of denials and

hardships due to the presence of only the psychiatric and cognitive symptoms. This leads to family abuse, sense of outbursts, severe depression, mood changes and, frankly, job loss.

So now I would like to turn it over to Dr. Andrew Feigin who will talk about our centers and what we would like to see done. Thank you.

DR. FEIGIN: Thank you, Barbara. Thank you to the Social Security Administration for holding these hearings and allowing the opportunity to prevent Huntington's Disease as potentially a disease that should be allowed under Compassionate Allowances.

So -- my name is Andy Feigin. I am the director of the HDSA Center of Excellence at North Shore Hospital on Long Island. The HDSA, in their wisdom, established these centers of excellence to provide multidisciplinary care to patients and

families with Huntington's Disease because Huntington's Disease is not just Huntington's Disease Chorea, as it used to be called. Huntington's Disease is a -- as Barbara mentioned, a multifaceted disease and pervasive disease that affects all aspects of a person's functioning, from their social functioning to their motor functioning to their psychological functioning, their cognitive functioning and, of course, all those things impact their ability to work.

So the centers of excellence not only have neurologists like myself, but we have psychiatrists and social workers and nutritionists and physical therapists and neuropsychologists. And -- but despite all of this, Huntington's Disease remains an inexorably progressive devastating disease that produces significant disability even very early in its course.

It is worth just reiterating something Barbara said, which is that there

are FDA treatments for the indication of Huntington's Disease to this day. We have medications to treat aspects of the disease, but, really, I would agree with the term that Barbara used, "limited," in the sense that depression is a common feature of Huntington's Disease. It was described by George Huntington in the 19th Century. Suicide is a common feature of the disease, an organic manifestation of the disease. And we do use antidepressants, with some success, but this is obviously something that is really just almost -- I wouldn't say it's a side issue, but it doesn't impact this inexorable progression of the disease.

I would also reiterate that I have many patients -- I take care of about 200 patients with Huntington's Disease, and I have been taking care of patients with Huntington's Disease for about 15 years. I have seen many patients who have applied for disability benefits and have been turned down.

In my opinion, actually, the diagnosis of Huntington's Disease should be an easy case. This is a truly devastating disease that, even early on, causes major disability in a person's functioning, and again, to emphasize it's really not just their motor functioning.

I think one of the major reasons that the people are turned down is the emphasis on motor disability. Huntington's Disease used to be called Huntington's Chorea. I think if you see somebody with Huntington's Disease, the thing that strikes you is their motor problem, their chorea. But from the perspective of people who take care of patients with Huntington's Disease, the chorea and the motor problems, although they are real, and people ultimately develop problems with walking and ambulation and end up bed-bound -- the major cause of disability in the early part Huntington's Disease is the cognitive -- the dementia and the psychiatric

disturbances. It could be as subtle as a personality change to as severe as frank psychosis with hallucinations, paranoia, delusions.

And that's really the major cause of disability, and we really don't have good treatment for it. Somebody who carries the diagnosis of Huntington's Disease, the clinical diagnosis of Huntington's Disease, is -- and is claiming disability, in my opinion, should be eligible for disability.

Having said that, I do want to talk about the issue of biomarkers and the issue about how the diagnosis of Huntington's Disease is made. I do research with imaging, PET imaging and MRI in Huntington's Disease. Barbara mentioned that Huntington's Disease is a -- is an autosomal dominant disorder so that every child of an affected individual has a 50-50 chance of inheriting the gene and getting the disease. It's a hundred percent penetrance.

The average person with Huntington's Disease gets the disease when they're about 40 -- 30 to 40 -- and then lives with the disease for 10 to 20 years. So most people with adult-onset Huntington's Disease function normally, go to school, you know, have professions, and then develop this devastating degenerative disease.

We know, from imaging studies, actually, that at the day that a person gets diagnosed -- this is, they meet some -- they cross some threshold of signs and symptoms that a doctor, looking at them, says, "you now have clinical Huntington's Disease's" -- they have had a degenerative disease for many years. We know that from imaging studies, looking at Dopamine receptors and imaging studies using MRI volumetrics. So people have significant disability even the day they get the diagnosis.

The obvious biomarker, I think, that people think about when you think about

an autosomal dominant disorder, we do genetic tests -- it's a commercially available genetic test. You can send someone's blood sample and get a genetic test that shows that they have the gene for Huntington's Disease.

This may sound like an obvious point, but I think for people who don't see genetic disorders very often, this may not be so obvious, that having the gene mutation for Huntington's Disease is not the same thing as having the disease. And I would not claim that everybody who has the gene mutation for Huntington's Disease actually meets the criteria for requiring disability, nor would the patients themselves, actually.

Most people who have the gene mutation for Huntington's Disease are -- you know, live totally normally, as I said already. They become -- I had a patient who was a commercial airline pilot. I had a patient who was a -- I had several patients who were doctors, you know, who go on and

achieve, you know, a high functioning in society and then develop these degenerative diseases. They, of course, had the mutation early on. They obviously were not disabled when they had the mutation.

The thing that defines the disability, in my opinion, is the clinical diagnosis of Huntington's Disease. If you have reached the point where your signs and symptoms were enough for a doctor to diagnose you with Huntington's Disease and you are applying for disability, I don't think anyone would argue with -- with your eligibility at that point.

I do want to reiterate one other point that Barbara made, which is that people often deny their signs and symptoms. So the -- that is a common feature of Huntington's Disease. I think it's, as I say, an organic feature of the disease, but it's also a psycho-social aspect of the disease.

Many of these people have grown up

with a parent with Huntington's Disease, an aunt or an uncle with Huntington's Disease, a brother or sister with Huntington's Disease, and they've seen what it has done to them in terms of their behavior, in terms of their appearance -- motor appearance with chorea. And it's actually -- it shouldn't be, but it causes shame and embarrassment. And they say to themselves, "I will never look like that." And their reaction, when they do start to look like that, is, "I don't look like that."

And so that can also have a major impact. I would say, actually, as far as disability is concerned, it's a positive impact in the sense that people will work until they -- you know, they absolutely totally are no longer able to work. I've never seen a patient inappropriately make a request for disability with Huntington's Disease. It's always the opposite, that people are obviously disabled and still trying to maintain their work, basically. So I'll

stop there.

MS. BOYLE: I would just like to say that we have centers of excellence around the United States, 21 of them that are -- that have directors with this knowledge of Huntington's Disease. And we would like to offer to the Social Security Administration information on how we could work with your facilities out there to determine and move this diagnosis process through for a positive ending.

What we did do is we left a -- a booklet for you and for members of your team so that -- that has the listing of all of our centers. And we would like to be able to offer our assistance and offer our knowledge to be able to take this process through so that our families don't end up getting lost in the system for a year or two.

And I would like to leave you with a message from one of our family -- one of -- the mothers of one of the people who are

dealing with Huntington's Disease. She says, "The signs most consider softer signs are minor movements, such as tapping of the feet and shrugging of shoulders, the beginning of chorea, but not the emotional problems which are the very first signs of Huntington's Disease, and those not recognized by most doctors across the United States, those that make it nearly impossible to live with our family members affected by HD and cause loss of jobs, divorces, loss of our friendships and social interactions."

So I thank you very much.

COMMISSIONER ASTRUE: Thank you. Again, we're going to hold off our questions until the end. We'll move to Pat and then we'll have questions.

MS. FURLONG: Thank you very much. I am Pat Furlong. I am from Parent Project Muscular Dystrophy, and because that's a mouthful to say, I like to just refer to us as PPMD.

We focus on Duchenne muscular dystrophy. Our mission is to improve the treatment, quality of life and long-term outlook for all individuals affected by Duchenne muscular dystrophy through research, advocacy, education and compassion. I am testifying on behalf of those families, but I do have personal experience in that my two sons were diagnosed with Duchenne muscular dystrophy. I am the de novo mutation in my family, and my two boys were diagnosed at the ages of four and six in 1984 and dead by the time they were 15 and 17.

So I appreciate the opportunity because we're thankfully seeing an era where this disease, once 100 percent fatal before the age of 20, is now moving into an area where the boys are living longer lives and they want them to be quite productive.

So there are several key points I would like to make in my testimony today. One is that there is confusion on the part of what

programs to apply for, and when, on behalf of our families. There is a lack of information, knowledge and categorization of these rare conditions within the systems and with those processing the claims. A lack of knowledge within a given disease-specific community related to what the programs might include, and concern that SSA programs discourage independence.

I think that this term "progressive illness" is very nebulous because people talk about Duchenne muscular dystrophy and muscular dystrophies as progressive diseases, but what does that mean in terms of progression? And it would be very useful if we had some sort of scale of progression because, often, the needs and services that are required at a certain time are really going to change, and really we can predict that they will change and what they will change to over time.

The exact time frame of those changes is more unpredictable. Muscular

dystrophy is a broad term that refers to a number of primary diseases of muscle. They include the dystrophinopathies, which are Duchenne and Becker, myotonic muscular dystrophy, distal myopathies, Emery-Dreifuss, facioscapulohumeral dystrophy, oculopharyngeal, and limb girdle muscular dystrophies, which are part of the dystrophinopathy group.

Many aspects of the diseases are not understood, and treatments are either unavailable or minimally effective. The age of onset and severity of the disease is highly variable.

You just need to mention Duchenne muscular dystrophy and you will be responded to by either, "oh, yes, I have an aunt or an uncle with MS," or, "I've never heard of this disease."

Most people ask about it, but once you see a boy walk with this disease, you'll never forget the image of a child with a

waddling gait and a child who's falling quite frequently. It's the world's most common and catastrophic form of genetic childhood disease. Duchenne represents 90 percent of all childhood-onset muscular dystrophy cases, and is characterized by rapidly progressive muscle weakness that results in impairment and weakness and often death in their 20s.

One in 3500 male children will be born with the disease, and about a third of them into families with no previous history of disease. The boys will lose their ability to walk in their teens. They will gradually lose the ability to move their arms in their late teens, and will have pulmonary and respiratory, cardiac and sometimes cognitive problems along the way.

So this is a multisystem disease. It affects everything these boys do, everything that they need to do over time. And just when they're developing the skills that they need to function, these skills

slowly melt away. And so they're left usually largely dependent on a wheelchair certainly by the age of 15, and unable to move their arms in their late teens with cardiac and respiratory needs as well, often ventilation during the day and certainly during the evening.

Individuals who apply for benefits for a child diagnosed with Duchenne muscular dystrophy are often applying for Medicaid or the Medicaid Waiver program through their local Department of Health/Family and Human Services. Many of these families are unaware of both programs, and many have no knowledge whatsoever.

The application process required in most states uses the same guidelines to meet the criteria for being disabled, and this results in confusion for families, as often they are unaware of the requirements to qualify as disabled, and it's often based on income levels.

In some states, approval is granted within two to four months, while in other states the approval process can be quite long. Long waits are the result of many factors, to include the family's unfamiliarity with the process. These families often fail to provide the necessary materials and have difficulty obtaining supportive medical documentation, to include an appropriate clinical diagnosis.

Clinical diagnosis and certainly genetic diagnosis is now easily available to these families.

Families are often unable to provide sufficient documentation to fulfill the criteria for being disabled. There is sometimes a significant difference of opinion and lack of evidence in order to determine what is medical necessity or what is educational necessity.

Individuals with catastrophic, disabling diagnosis want and deserve to be independent, and developing systems that

provide benefit while encouraging their independence is essential. Individuals who are able to work, even part-time, should not be denied benefits; thus, this would discourage their independence.

I really polled some of our older patients -- I never know what to call them -- if I call them old guys or young guys or, certainly, adults with Duchenne -- and I asked them what they would like me to tell you.

There are several things that are really on their minds these days. One is, in all medical literature, this diagnosis is described as 100 percent fatal. They relate this as people processing claims, people having knowledge of the disease to someone with a gun to their head and someone ready to pull the trigger when actually they, in their terms, will tell me, it's hard to die well over 20 years, so let's stop calling the disease fatal, and let's have a definition of terms that clearly reflects the disabling

condition as opposed to a fatal condition.

They would like a scale for rates of progression because often they will apply for needs, and these needs change over time -- they change in some boys quite quickly.

They think there should be a tutorial for applicants, especially as they grow older and they are adult and want to be independent. They would like to know how to apply, what to apply for, what they might ask for, what they might not ask for, might not be able to apply for.

These young men are really interested, with use of technology, to become independent. So they really are -- feel like they're discouraged from being independent because if they receive benefits, that they can't be a wage earner, and they're not often able to really earn sufficient wages to cover all of their needs.

So they get into this dilemma of the sort of chicken and egg. If they work and

become productive, as they want to be, they lose benefits, and they can't possibly work long enough to have as significant of a wage to cover their benefits.

So their really final word was to encourage you to develop a mechanism by which they can maintain independence for as long as possible and they can be productive individuals with using technology, at the same time, to be able, once that need has changed, to receive their benefits once again, or to continue their benefits along the way. Thank you.

COMMISSIONER ASTRUE: Thank you. I have a bunch of questions. I'm trying to figure out how to organize them.

Let me start with a couple of questions on Huntington's. So, typically, medical advances make our work easier, rather than harder. It may, in the case of Huntington's, make it a little bit harder because I think if this were 1965, usually by

the time someone was definitively diagnosed, they would have progressed fairly far and there wouldn't be too much question that they would be disabled under our standards. I know that misdiagnosis was very common for a long time.

Now, where an increasing number of people know, you know, at a very early age what they're likely to be facing, we have to figure out where to -- under the statute, we have to figure out the appropriate place to draw the line because, if the disease is going to start making substantial impacts sometime between the age of 30 and 45, the statute doesn't allow us to give benefits at age 25.

So we need to figure out sort of how to draw that line. I mean, I think as long as we can draw that line effectively, it strikes me that in the case of this particular disease, most of the rest of this is fairly easy because I think, you know, you don't have a range of severity and that type of thing.

Can you talk to me a little bit more, Dr. Feigin, about sort of what would you -- what do you use clinically to make a definitive diagnosis that someone really is, at that point, symptomatic and starting an actual progression?

DR. FEIGIN: Yes. I wish I could say that there is, you know, a total score on some scale that, above that score, somebody gets a diagnosis of Huntington's Disease and, below that score, somebody does not get a diagnosis of Huntington's Disease. Unfortunately, that is not the case. There is no consensus about what degree of combination of motor, cognitive and psychiatric signs and symptoms you have to have in order to achieve a diagnosis of Huntington's Disease.

And probably even among Huntington's Disease experts there is some variability in that in the people who are in that kind of few years' period where they are hovering, you know, between, well, do you call

them having Huntington's Disease or do you say that they're not quite there yet? Some doctors would say, "I'm going to call that person a Huntington's Disease," and others would not.

There is a point, though -- and it's not well-defined, unfortunately, at this point, where everybody would say, "That person has Huntington's Disease."

And I guess I would say, even though that is not an extremely well-defined point, that is the point when people should be eligible for disability.

I'm not sure that -- I would say I am sure that no consensus has been reached on exactly when that point is, but I do think that one aspect of it -- I guess this is not something that is necessarily reliable, but is -- I just -- I think that -- let me just take a step back. I do think there are a lot of people who have the diagnosis missed for a long period of time, and when they do come to

one the centers, or they go to somebody who has some experience with Huntington's Disease, they clearly are at that level you were talking about where they are disabled.

I do think for people like you're describing who know they're at risk and are going to an experienced center who -- and are being scrutinized over a period -- you know, longitudinally, you know, every six months: Do they have eye movement abnormalities? Do they have other things? You know, extensive neuropsych testing. There is a gray zone, I have to agree, and I don't know if I have an answer to that. But I do think that that's a brief period for people with Huntington's Disease, and then they go into the phase where nobody would argue about it.

COMMISSIONER ASTRUE: In trying to figure out where to draw a line, does age help us at all? I mean, is the natural history sufficiently consistent that you can say with a fair degree of confidence that the disease

doesn't really -- is not really symptomatic before age 30? Or is there enough variation in the population that we couldn't say that?

DR. FEIGIN: Yes, I would say that there is enough variation that you can't really say that. You know, having said that, there are statistical models based on the mutations. Based on the mutation, the CAG repeat, the triplicate repeat, which is what causes the Huntington's Disease, that based on a person's age and CAG repeat length, you can predict within some range of probability when they are going to start showing signs of Huntington's Disease, that would mean a diagnosis of Huntington's -- that's based on thousands of patients who have been diagnosed, and knowing their CAG repeat length and their age at the time that they were diagnosed, and then creating, essentially, life tables -- and that's published, actually -- life tables based on CAG repeat length and age at any given time in life and probability of showing

signs by a -- a sign sufficient to diagnose Huntington's Disease by a given age.

So there is information out there. The problem is -- it's a long-winded answer -- there is a lot -- you know, there's a lot of variability around that. There are juvenile-onset cases of Huntington's Disease. The earliest purported case was the age of two. And so -- the latest is in their 80s, so it's very -- it is, unfortunately, very variable.

The juvenile-onset cases are, as you might guess from what I just said, are the cases that are associated with these very long CAG repeat expansions, so...

COMMISSIONER ASTRUE: I mean, I think, you know, the general principle that -- I mean, this is a disease that we ought to be giving accelerated condition of. I don't have any disagreement with that at all. I think the question for us is just, you know, how to draw the line in an efficient and fair way

that's consistent with the statute.

Any guidance based on clinical experience at these centers of excellence or anything else you can do to help us figure out how to draw that line in a way that makes the most sense, consistent with the statute, that would be greatly appreciated.

MS. BOYLE: I think the interesting thing is that only 10 percent -- a maximum of 10 percent of our families at risk for Huntington's even go and get the genetic test taken. So they live on the line that they're not going to get this disease.

And I have to tell you that most of the times when they go to the center, we have an expert, a neurologist, who understands Huntington's. And when they give them a clinical diagnose of Huntington's, they are well into Huntington's Disease. And it may not be the chorea, but it is abuse in the family, it's paranoia and sitting in a room, never leaving that room for four and five and

six months.

So I think if we could work with you with our centers of excellence and with our group of neurologists, who are very, very willing to work with every member and guide them on clinical diagnosis, then we would be certainly establishing a precedent for moving forward.

COMMISSIONER ASTRUE: That's helpful.

Let me move over to CF for a second. As we try to figure out subpop -- I mean, obviously CF is very complicated because it is multisymptom and there's a range in severity and we can't just give out benefits to everybody with CF, and we've just got to figure out the most intelligent way to draw lines.

Is there -- any of the symptoms -- any of the other conditions that are associated with CF that, in combination, would be something that we ought to be looking at?

For instance, is there anything -- when you've got -- is it inflammation of the pancreas?

DR. BOYLE: It's actually insufficiency; they tend not to have inflammation.

COMMISSIONER ASTRUE: Insufficiency? I mean, is there -- are there a category of cases where a problem with the pancreas in combination with the other problems with CF -- you would be able to look at that and say, with a degree of confidence, that you've got that -- that combination of impairments; you ought to draw the line there? Or is that still too diffuse?

DR. BOYLE: Unfortunately not for pancreas because about 85 percent to 90 percent of individuals are going to have pancreatic insufficiency and there can be a spectrum of severity. So -- I know what you're asking. You're trying to say, are there other things we can look at that will help tell us about the severity of what's

going on?

Unfortunately, the genotype -- I believe I mentioned this before, but genotype doesn't tell us. And we know that about 50 percent of all individuals with CF have the same CF genotype, and there is a mixed spectrum of disease with that.

So genotype tells us, "yes, you have CF," but I don't think it helps answer that question that, to me, the biggest -- there are a couple of things -- poor nutrition can be one of the ones. Sometimes it's liver disease, although that's fairly rare.

To me, the biggest thing that helps is trying to look at the frequency of illness and the burden of care, how much time there -- because those are the things that I experience which have the biggest influence on whether or not that person can work.

And I will say -- this is, actually, a flip side -- I'm usually telling our patients, "hey, you can do this."

Most of them want to work. I spend all my time saying, you know, "you're going to live to be older," so this is a little bit of the flip side. But two ways, I think, that we can measure that best in terms of burden is looking at the acceleration of their care. And so when you look at what medicines they're on, early on, they tend to be on some mucolytics, things that break up mucous, and that doesn't really reflect the severity of the illness, but as they get inhaled antibiotics, and they're doing that more frequently, every other month twice a day, that's a real sign of --

COMMISSIONER ASTRUE: Is there just one out there now, or are there multiple inhaled antibiotics?

DR. BOYLE: There are two that are used and another one which should be approved for use in the next year. And I think what you're actually going to see is more and more -- more and more burden in terms of

trying to do these medications. But I think that's actually a pretty good survey because the recommendations for use of inhaled tobramycin, which is an FEV1 that's below 75 percent of predicted -- and we tend to use a little more infrequently until the patients are, obviously, showing more problems, and we keep adding it on till they're doing it every other month.

The other one is just the frequency that they're ill. So exacerbations, as judged by -- hospitalization is an easy one, but I think the home IV antibiotics is another one, the number of times that they are requiring some other intervention, to me, those are the things which add up which make it hard.

COMMISSIONER ASTRUE: I agree with that. I think the physician intervention, I'm always sort of dubious about that. It seems to be not a very good indicator -- hospitalization or, you know, hooked up to an IV, I think we'd all be a lot more comfortable

with that.

DR. BOYLE: Suzanne may have a comment, but I think the way things are written right now actually are -- they are written well because they actually do appreciate that there's not just a number that tells you how the person is doing. I think sometimes it's how they are acted upon. Because it's almost like you have to have all of the above rather than, you know, if your FEV1 is not horrendous and you're ready for a lung transplant, there can be certainly a lot of other medical -- you can be really sick. You can have an FEV1 of 70 percent and be in and out of the hospital or you're doing home IVs a couple of times a year and struggling to function.

COMMISSIONER ASTRUE: Let me ask a question for Suzanne. I mean, sometimes it's both a good thing and a bad thing when -- dealing with us when you're multisystem. I mean, sometimes it is confusing for people

that have to work their way through the system, or the people that administer the system. But sometimes it does mean you have multiple bites at the apple. So we are -- as you admit, we are working pretty hard to come out with some new -- the new respiratory listings, and we do appreciate what you can put into it.

We have just come out with new digestive guidelines which will be effective in just a couple of weeks, and I just wanted to ask you if you had any input on section 508 which theoretically, I think, CF patients could use the new BMI index as a way of meeting the listings.

Do you have any -- maybe you want to look at this and supply it for the record, but do you have any thoughts on 508 and the digestive regs, whether we drew the line at the right place with regard to CF patients or whether you're fairly happy with that.

MS. PATTEE: I have to confess that

we did not actually contribute to the digestive listings. We missed those. We were focusing on respiratory. But when we commented on the respiratory, we commented on the growth impairment for children and felt those were too severe.

And then when we sent in the letter for Compassionate Allowance, we encouraged a specific BMI level being looked at as well. I think it was 10 percent. Now, maybe I could pull out these numbers that I don't remember, but I can certainly get back to you and specify what our recommendations are for the digestive.

COMMISSIONER ASTRUE: Okay. I think that would be helpful, and I think it probably also -- you know, it may be that we will have something that's more helpful when we go through the respiratory listings, but at least this is -- might be a helpful change for some people, so we should probably have some discussion about it, and it might also be

helpful to do little bit of outreach in the patient population and -- this is another thing that they may be able to rely upon.

MS. PATTEE: We appreciate the look at -- the use of BMI because we thought that was missed on previous things.

COMMISSIONER ASTRUE: Good.

Multiple [sic] dystrophy.

I wonder if you could talk to me a little bit more, if you can, about the patients that experience some difficulty going through our system. Do you have a sense that that's because of confusion about the disease or do you have anything else that you can give me little bit of a handle on as to, for those cases, why they're going off track?

MS. FURLONG: I think some of these cases are going off the track for a couple of reasons. One is this is a complex multisystem disorder, so in addition to progressive muscle function loss, some of these individuals have some issues related to respiratory and

cognitive, so some of these children have learning disorders and they run through the range of simple learning disorders or processing disorders through autism. So some of the boys have severe mental loss of function.

So I think understanding where this child fits in the spectrum and what services that they might use or access is difficult for the parents to understand. So I think that that's been the difficulty as well as there has been some differences in Medicare and Medicaid Waiver across states where some patients are waiting for very short periods of time and other patients find that they're on a waiting list.

So there has been a range of issues from these patients and parents.

COMMISSIONER ASTRUE: The other question I wanted to ask -- and I know my staff tends to cringe when I say things like this, but I'll say it anyway -- I mean, it

seems to me that a lot of the issues here are probably substantially simpler for children -- and we have a related, but different, standard for children -- but potentially more complex for the adult population for some of the reasons you started to touch upon.

Could you talk to me a little bit more about sort of what percentage of the overall patients are adults and what their lives tend to look like and what your sense is of how many of them try to work and what type of work and what persistence of the work effort is, and just give me a little bit more touch and feel on that because I think that would be helpful, at least to me.

MS. FURLONG: Sure. I don't think we have accurate statistics today to tell you about how many are interested or willing to enter the work force. I can say that because the disease is changing, based on care and based on longer lives and cardio protection, that these boys are living into their 20s and

30s. There are single cases now of young men going through college and on to having careers. But this is more and more becoming acceptable and routine for families so that they're not assuming that this boy is going to die before 20 and not be able to live to be an adult.

So I think that, over time, then, we're seeing boys enter the work force, but it's not as simple as entering the work force and becoming a productive individual in terms of they have a loss of muscle function, so they need jobs in technology. Also, they need someone to help them, you know, do the activities of daily living because they aren't capable or don't have the function to be able to take care of themselves in terms of toileting activities, daily living related to combing your hair, transportation and so on.

So it's that dilemma where, you know, you would like to fit them in a box that says that they can have a job and be

productive, but in order to achieve that, they're going to need some services available to them, and then they don't have the endurance to work full-time, for instance.

So there is that very difficult spot where the loss of services is significant, and the ability to work and generate an income sufficient to cover your needs is impossible. So it is a very dicey area that we're just seeing.

COMMISSIONER ASTRUE: I may have more questions now -- I've got a tangle of note here, but let me try not to hog the forum too much. David, do you have any questions?

DR. ECKSTEIN: The role of the Office of Rare Diseases is a little bit different than -- it's sort of a hybrid between what the applicants are doing in providing advice and what social security is doing in actually making regulations, so we're a little bit more involved in that. But there are other activities that we can do that

ultimately can impact the decisions here.

And so I have, actually, a question for Dr. Feigin regarding the sort of standardization of the diagnosis. You had indicated that it's fairly variable right now. But is the community ripe, is the field ripe for bringing together the people to make a standardized type of diagnosis that would then be the standard to which this could be reflected to, such as care.

DR. FEIGIN: Well, the answer to that question is certainly yes. There are organizations of people, like-minded people interested in improving care and finding new therapies for Huntington's Disease, like the Huntington's Study Group, for example, that involved hundreds of people from -- you know, people like myself from around the world, actually, that I think would be interested, potentially, in trying to do that.

I think, though, that the -- it would be a difficult task because there is

this area in which -- where patients, you know, have the gene for Huntington's Disease enter where they have very -- where they enter this kind of gray area where I think there's going to always be differences of opinion about how much weight to give to those things.

But I'm glad you asked me the question because I felt like after -- there was another point I wanted to make, actually, related to this issue, which is I mentioned that if you knew you were at risk for Huntington's Disease and you were going to a center and being scrutinized every six months or every year, you know, that would present you with this problem. But that represents a tiny fraction of people, I would say, who are at risk for Huntington's. The vast majority of people don't go to a doctor at all, actually, and they're not in this situation where they're being scrutinized.

For the people who are doing that, it's actually not the people that enter the

this gray zone, or even people in the very early stages of the -- of unequivocal diagnosis of Huntington's Disease, that first year or two. Those people are not applying for disability, actually, in my experience. They are not -- it's the people who have crossed that threshold to the point where they clearly have signs of Huntington's Disease who are the ones -- and even they often are not applying. It's their families that are pushing them to do it.

It's really -- we're talking about a very small fraction of the people who -- of a small fraction of -- a small fraction of a small fraction who this discussion would even be relevant to, actually. The vast majority of people who are -- with Huntington's Disease who are applying for disability are in that phase of the disease where nobody would argue about the diagnosis, so...

JUDGE CRISTAUDO: Ms. Furlong, do you have a sense if individuals with the

Duchenne impairment, do they reach the hearing level in our process or are they generally approved before it reaches the hearing level?

MS. FURLONG: In general, they are approved before it reaches the hearing level.

JUDGE CRISTAUDO: Some get to the hearing level?

MS. FURLONG: Some get to the hearing level.

JUDGE CRISTAUDO: And I believe with the other two conditions they do actually reach the hearing level, I am to understand --

(Nodding heads.)

JUDGE CRISTAUDO: Once it's at the hearing level, are most of the cases approved? I'm sensing from Dr. Feigin no.

DR. FEIGIN: No, no, no. I think it's yes, actually.

JUDGE CRISTAUDO: Most are approved?

DR. FEIGIN: Yeah, but it's the frustration -- you'll actually read some

attestations in here from patients that I think reflect my experience, too, which are the people go through the application process; it can drag on for a long period of time. They walk in for their hearing, and it's a no-brainer, which once the person actually sees the patient and talks to them for a couple of minutes, they realize that the person is disabled.

JUDGE CRISTAUDO: Is that your experience also, Ms. Furlong?

MS. FURLONG: Yes.

JUDGE CRISTAUDO: And also --

MS. PATTEE: Yes.

JUDGE CRISTAUDO: So then -- are most people who have all of the conditions that you're representing here today, are most of them treated by specialists?

MS. PATTEE: That is the case in cystic fibrosis, yes. We have 120 care centers around the country.

JUDGE CRISTAUDO: And then you're

saying, with Huntington's, there are generally generalists --

MS. BOYLE: No. In our centers, we probably see about 5,000, and there are 30,000 active HD patients. The problem is that by the time they apply for disability, they really are into the system -- into the process of having real psychological and emotional problems.

If they have chorea, that's a different thing. Then they are willing to see that and admit it. But it's the psychological and emotional problems that they have that have caused real damage. I mean, we have probably about 5 percent of our patients that go through the jail system before they're even -- before we're even able to see them and we're able to be able to work with them to get them on disability and to get them the kind of help that they need. So, no.

JUDGE CRISTAUDO: But the people that actually go to the hearing, are they

treated generally by a specialist?

MS. BOYLE: Yes, I would say so. A good part of them eventually, at that point in the game, after they have been turned down a few times and then they end up going to the hearing, they've called us at our office, and we then try to get them to a center at that point in the game, yes. But sometimes it takes a year or two.

JUDGE CRISTAUDO: Sure, I understand.

And Ms. Furlong, same thing. I mean, generally, if they get to the hearing level, they're treated by a specialist?

MS. FURLONG: Most of the time they're diagnosed certainly by a specialist, but there is a certain percentage of families that are lost to follow-up based on the message that there are few interventions that would change the course of progression. So there is a -- probably 25 percent of those patients that are then are treated by their

family doctor or their pediatrician, rather than a specialist.

JUDGE CRISTAUDO: Okay. And then my final question is a follow-up on something I've been trying to pursue, is -- and we have two doctors on this panel, of course -- it sounds like that some of these cases are getting to the hearing level, they get to the hearing level, and it's pretty automatic, is what I'm sensing.

So what I'm trying to figure out here is what do we need to be asking the doctors differently, what do we need to be doing with the doctors earlier in the process so that they actually produce the information that we need so we can allow the case that's going to be allowed anyway?

If either one -- or any of you, actually -- the two doctors in particular -- if you can respond, I would certainly appreciate it.

DR. FEIGIN: I mean, for

Huntington's Disease, I think the thing that I think would be important, actually, would be -- as I mentioned, I think, earlier, that there is an emphasis in Huntington's -- you know, Huntington's Disease used to be called Huntington's Chorea. There is this kind of perception out there that this is a movement disorder. I am a movement disorder neurologist. I trained in movement disorders, and I take care of people with Huntington's Disease.

But there are Huntington's Disease centers around the country that are a lot less arduous [phonetic], actually, and I think that there needs to be more -- I think the information that should be requested and should be provided that might make the difference is cognitive and psychiatric information, that more -- in addition to the motor information, yeah, in the case of Huntington's Disease. If that means just the exam or if it means things like

neuropsychological testing or if it means things like a DSM-IV diagnosis -- I mean, all of those things. I couldn't go into it.

DR. BOYLE: Actually, I think there's two parts to the equation for CF. One part is on the hearing side and one part is on the physician side.

I think oftentimes experience has been that the initial review is hesitant to look at much more than the FEV1 because everyone is so used to thinking of cystic fibrosis as a lung disease and so you think, "oh, it's almost a variation of COPD."

They look at the FEV1 and if it's not that bad, they say, "you know, you're not really that sick. Okay. We'll hear a little bit more of the details later."

And that's one reason why I think we focus somewhat on the idea of, "okay, let's look at these other criteria as well." And I think one of the things that might help clarify that would be perhaps providing a few

more practical examples of what these things mean in terms of the criteria.

And then, also, it's on the physician side, too, in that -- some of that same issue with thinking of it's all about FEV1. But one thing I want to say is that the Cystic Fibrosis Foundation has really done a good job of trying to educate the CF specialists about this. And hopefully it will be something that will be improving. I just want to make sure that once the physicians get with the program and are making sure they're supplying the right things, that there is also the understanding on the other side of the table.

And so specific things to ask about are things like the frequency of inhaled antibiotics, the frequency of home IVs, not just -- my sense is FEV1 and hospitalizations sort of rule the day for the course of time.

MS. FURLONG: I think -- just to make a comment about the muscular dystrophies,

because they are a group of disorders, I think there is confusion in what those -- each of the disorders represent, because there's -- age of onset is different, clinical depression is different. So to have really concise definitions of what those are and what the rates of progression are in each of those indications would be very useful, because then you can put those in a box that is not always going to be a clear, well-defined box, but at least it's a more understandable box.

COMMISSIONER ASTRUE: And that's a perfect setup to what, I think, may very well be our last question before the lunch break.

So Duchenne I'm relatively familiar with, and one of the key things here I think there is some reason for hope over the long run, because I've seen what a couple of really interesting small companies are doing in the area -- and I can't talk about it because I signed a confidentiality agreement, but there's at lest reason for hope, and that's

great, and it's something I'm generally familiar with --

MS. FURLONG: The landscape is changing dramatically.

COMMISSIONER ASTRUE: Things are starting to change, and that's great.

But I'm really very ignorant about the other dystrophies and the other related diseases. And I wondered if you could help me a little bit to understand -- if we want to get a handle -- a better handle on those -- and I'm sure we've got people somewhere in the agency who do have a better handle, but at least for me, does your group represent those? Are you focused really on Duchenne or do you represent all the other related diseases?

Are there other groups and centers of medical excellence we ought to be aware of? I'm just trying to figure out, to the extent that there is a network in this whole disease area, I just want to make sure we don't miss anybody.

MS. FURLONG: Yes. The Muscular Dystrophy Association represents the muscular dystrophies as a group, but there are also disease-specific groups like myotonic and limb girdle muscular dystrophies under those, so there are disease-specific as well as the large umbrella organization, which is the Muscular Dystrophy Association, and they certainly represent these diseases as well.

COMMISSIONER ASTRUE: That's helpful. Anything else?

Okay. I think that we're pretty much done. I have to do an apology again today. We are in a -- close to a restaurant-free zone done here. That may be changing with the renovation of the building that's diagonally across the street from us. But for now -- it's the reason we've given a longer lunch break.

Please, everybody, be especially careful today. We have had a little bit of rain and wet snow out there, so I'm sure it's

very slippery. If any of you -- there should be a restaurant guide in your packets, but if you don't have that, just go over to Diane Braunstein here on my right, and she'll be happy to help you out.

Again, this is another terrific panel. This is extremely helpful to us, and we are exceptionally grateful for your contribution here today. So thank you.

(Lunch recess.)

AFTERNOON SESSION

COMMISSIONER ASTRUE: We're going into our third and last panel before we go into general and public comments, and we're very pleased to welcome Shelley Bowen, president of the Barth Syndrome Foundation, Josephine Grima, the vice president of research and legislative affairs for the National Marfan Foundation, and Amy Kirk, coordinator of family services for the Batten Disease Support and Research Association.

Do you have a preference as to who goes first?

MS. BOWEN: I'll go first.

First of all, I would like to thank the organizers for inviting us to come here, and it's an honor to be able to speak on behalf of the children who have Barth syndrome.

Barth syndrome is an X-linked disorder. It was -- until the mid-1990s, it was considered uniformly fatal in infants. My

experience with Barth syndrome was as a mother. I was a de novo carrier, just as Pat Furlong was, of two boys who had the disorder. One of my sons died in 1990 from Barth syndrome, and I now have a 20 -- will be 21 on Friday, who has Barth. So he is a new survivor, but with that comes new and unique challenges.

We have had some experience personally with -- some challenges with Social Security with getting Michael listed as being disabled, and I went into that in my written testimony. And at this point I'm hoping -- I am kind of sad that I was so worried, this whole description -- but what -- to go to -- I would just like to describe a bit about the disorder.

It's a severe inherited error in metabolism. It causes a remodeling of the cardiolipin which affects the mitochondrial organelle of every cell of the body. It causes cardiomyopathy. You can have rebounds

of heart failure where children can go in and out of heart failure, fatal arrhythmias, skeletal myopathy, delayed growth. You'll see osteopenia and osteoporosis. And one of the aspects of the disorder that really isn't fully understood fatigue and weakness that these boys experience, sometimes being so dramatic that it's debilitating.

These rebounding issues of heart failure you will see -- you will see arrhythmias appear usually in the years of puberty, and the children -- the oldest children that we're aware of in the United States are into the second decade of life, about 25 years of age.

So these parents -- about 70 boys are in the United States. These parents have been very high-functioning families. They are just now beginning to see these issues where these children are growing up and getting out of school, from high school, and they're going into college. Most of them have been

home-schooled. The parents have had to adapt. These children don't have a skeletal muscular dystropy. They've never had muscles. They just -- it's always -- they've had -- they've never had muscle tone. So it might look like a beefy little child that's fat and looks very healthy, but the cruelest part about this disorder is that these children do appear deceptively healthy.

My greatest challenge with my son -- and I think I need to speak to this because Michael is one of the oldest survivors -- is that we had a challenge -- it took us nearly a year to get through the process with Social Security to get Michael listed as being disabled. It took a lot for me to check that box off to say that my kid that I trained and taught to be an asset to society -- taught him to be able, and then have to check it off. That was hard.

But realizing that he was no longer going to be qualified as a dependent on my

husband's insurance, I knew that I had to do something. So I spent -- the night before my son went into the hospital when he was in heart failure, I spent the evening going over the paperwork, and finally submitted it all, including the papers that describe the disorder, the gene discovery, all of the journal articles that Michael had actually been in with circles and tabulations of everything that would actually give the proof and the evidence that we would need for him to be considered disabled.

I was shocked when I received the paperwork back nine months later that said that while it's apparent that you do have a disability -- you state that are you disabled -- however, judging by your age and your education -- he was a straight A student. Granted, he didn't graduate until he was 20 -- you should be able to work.

He couldn't. He can't. He can only go to school four hours a week. And

that's Barth syndrome. He's bright, but -- he wants to do something. And unfortunately, now today, my son with Barth syndrome is uninsured. We have no coverage for him, and he's at risk.

So that's my experience, and unfortunately, I feel that there's a lot of people who are going to be behind me that are going to experience the same issues because the adjudicators are not familiar with the disease. Even the medicine society, really -- or the medical profession isn't that familiar with it because it is a complex multisystem disorder that affects every single cell in the body. It can be debilitating from very early on, and it can start -- and you can also see it slowly progress during the child's -- into the second decade of life.

So I would recommend -- if someone could help high-functioning families and help teach us how to advocate for our families and give you what you need. We do have a

biochemical marker that is being -- that has been developed -- right now, it's in research -- to distinguish if there is cardiolipin remodel -- or cardiolipin deficiency. This is distinct from the genetic mutation or looking at the tafazzin gene. So as was mentioned earlier today, not all of the children are going to be as severely affected as some.

But the cardiolipin deficiency would prove -- give you the evidence that you needed.

COMMISSIONER ASTRUE: I've got a whole bunch of questions, but I think we're going to stick to what we've been doing. We'll hear from everyone, and then that will give you all a chance to chime in together when we have questions that overlap in areas of interest. Thank you.

DR. GRIMA: My name is Josephine Grima and I'm the vice president of research and legislative affairs of the National Marfan

Foundation. And I'm honored to appear here before you today.

I would like to send a special thank you to Diane Dorman, even though she's not here, for her support and her intention to our concerns. And even though he doesn't want to hear it, I want to send a thank you to Dr. Stephen Groft who has been a valued friend of the National Marfan Foundation for many years.

And, lastly, I would like to thank the Social Security Administration for their leadership over the past year for producing an informational video on Marfan syndrome for their adjudicators so they can help better understand the syndrome.

Marfan syndrome is a rare medical condition affecting 40,000 people in the United States. It manifests itself in several body systems, including the cardiovascular, ocular, musculoskeletal, pulmonary and nervous systems and, to a lesser extent, the skin.

Marfan syndrome is a progressive disorder with virtually no prophylactic therapies to modulate the deterioration of the lungs, the joints, the eyes and the blood vessels.

The good news is that the life expectancy of Marfan patients has increased from 45 years of age to 70 years of age, primarily due to elective surgery, surgical repair of aortic roots. However, the aging Marfan population is a new entity, and the natural history of this older generation is showing complex disability issues resulting from multiple aortic surgeries, degeneration of bones and joints, visual problems associated with the dislocation of lenses, dural ectasia, resulting in back pain, headaches and difficulty moving their lower extremities, pulmonary problems, hernias and early onset of arthritis.

This is evident -- this is evident to us with the dramatic increase in inquiries

to our office during the past ten years. The chronic debilitating effects of this syndrome have become the leading factors of disability.

Individuals are unable to sit or stand for long periods of time to perform basic sedentary work activities. In addition, jobs with more physical requirements are not advised for people with Marfan syndrome because any strenuous activity can cause a dissection of the aorta or aggravate an existing aneurysm or dissection.

Even in closely monitored patients, instability of the aorta remains problematic, and it is a fact that Marfan syndrome is probably underappreciated by many doctors who do not see many Marfan patients.

Marfan syndrome is a life-threatening disorder, due to the potential for dissection of the aorta. Currently, Marfan syndrome is listed by SSA as a cardiovascular impairment specific to patients with dissections that are not

controlled by prescribed treatment. However, these uncontrolled dissections are the most severe of circumstances which, unfortunately, leads to loss of life in many instances.

Most dissections, although severe and disabling, can be controlled with medical therapies. The fact that cardiovascular disability determination is based on whether the dissection was uncontrolled misses the mark as the true indicator of disability, in our opinion.

In response to the questions posed to us, processing of claims for patients with Marfan syndrome who meet the disability criteria take a long time to be processed. Most people are denied on the first application and must appeal the initial decision, often multiple times.

For those denied multiple times, we believe this is due to the severity of the standards listed in the cardiovascular impairments. Marfan syndrome is a rare

disease and it is not well-known. The effect of the syndrome on the whole person and not just the cardiovascular system is not understood by the adjudicators or reflected in the listings.

Many appeals go all the way to the final judicial review. In many cases, Marfan patients have to be evaluated for effects in multiple body systems. They may not meet the severity retirements of any one of the body systems in other listings, but when multiple, less severe impairments occur, a disabled state can also occur as a result of cumulative effects across the body systems.

Adjudicators and, in some cases, physicians are unfamiliar with the multiple-system effect of Marfan syndrome and the effect it has on the day-to-day functional abilities of many people.

Another major problem is that physicians are unfamiliar with the Social Security application process and do not

include the important facts in the letters of support for patients. In addition, published medical evidence for many of the functional limitations is lacking. Therefore, physician requests for disability should be evaluated by adjudicators, and physician reports may need to be weighted more heavily in the evaluation process if an appropriate decision is to be made.

Finally, a lot of the SSA staff are unfamiliar with rare diseases and don't take the time with patients to listen and learn in order to better understand their functional disability.

Marfan syndrome cannot be expedited under terminal illnesses because it is not considered terminal. However, we were very aware of cases where people have had problematic descending aortic dissections who are denied disability, struggle to continue working, and die before ever receiving disability. Unfortunately, it is the lack of

appreciation of the instability of the Marfan aorta that appears to create this devastating result.

The supplemental security income program is definitely a good program for those that do not qualify for disability, but again, it has to have more information on rare disorders.

In order to improve the current system, our recommendations are as follows: A listing of rare disorders and their disabling symptoms and the impact on each body system for the adjudicators to use would be extremely helpful.

Multisystem disorders, such as Marfan syndrome, should be recognized as such so that the impact of multiple disabilities can be better recognized and evaluated.

Guidelines to evaluate multiple factors, which do not have concrete medical evidence, such as fatigue, need to be developed.

Guidelines for doctors that explain the types of information and medical records needed to document disability and helpful content for letters would also be useful.

Adjudicators also should have a better understanding of how difficult it may be for an individual for a rare disorder to obtain documents to support their application and, therefore, they should be more willing to assist patients with the application and obtaining those supporting documents.

When people are denied disability, many of them give up and do not understand the role of the appeal. This needs to be clearly stated in plain language in a prominent place. The reasons for the denial need to be specific and clear. And if the application lacks important information, this needs to be stated. The denial letter should explain what additional information might make a different decision.

In many cases, we see people

applying for disability who have no income. They lack medical insurance to receive medical care, which would address their medical needs and document their disability. So when applying for disability, many need to see SSA doctors for information to help substantiate their claim, but many of the doctors do not know Marfan syndrome. Having physicians that know about rare diseases would be a benefit.

Thank you, again, for this opportunity.

COMMISSIONER ASTRUE: Thank you.

Amy.

MS. KIRK: My name is Amy Kirk. I am the coordinator of family services for the Batten Disease Support and Research Association. And what I do is I try to help families get things like social security, Medicaid, help them out in the schools, so I work with the families to help them go through a lot of this process.

Batten Disease is the common name

for a group of diseases known as neuronal ceroid lipofuscinoses, and is one of the more common of the neurodegenerative diseases. It is also one of the diseases found in a group known as lysosomal storage disorders. It is a recessive inherited disease, meaning that both parents must carry the defective gene.

Batten Disease is rarely diagnosed immediately, and is often mistaken for epilepsy, mental retardation, retinitis pigmentosa, ADHD and even schizophrenia in adults. Onset is characterized by beginning vision loss, seizures, clumsiness, personality and behavior changes.

Batten Disease causes continual physical and mental deterioration, leading to death. Depending on the form of the disease, children may die as young as two or three, or may live into their 20s or 30s. At this time, there is neither a treatment nor a cure.

And many of our families have had to individually list out the conditions of

their child's disease when applying for social security benefits. We found that it's easier to have them list out every single condition of Batten Disease that their child is experiencing, rather than putting Batten Disease or neuronal ceroid lipofuscinoses on their application. When the average person, even the average medical professional, hears the term "Batten Disease," they have no idea what this entails.

To ensure that a family does not experience a prolonged delay in Social Security's decisions, we tell tour families to explain the symptoms of the disease as a means of diagnosis. When an SSA adjudicator reads seizure disorder, blindness, fine motor skill impairment, gross motor skill impairment, mobility disorder, et cetera, a quick decision from SSA is much more likely.

However, when the term "Batten Disease" is listed, there is always an explanation that needs to come along with it.

And for both the sake of our families and the Social Security Administration, we have recommended the individual symptom listing as the way to describe the diagnosis.

This, in itself, we have found poses two problems. One, it's not a true diagnosis. Yes, a child has a mobility disorder, but the mobility disorder is a condition of Batten Disease, and while we are not lying on the application, we're not really presenting the entire picture either.

The second problem is that providing medical information for each of these individual symptoms can be tedious to be all involved. Providing the Social Security Administration with a diagnosis of Batten Disease would make the process much easier and faster.

Instead of searching for ophthalmology records, physical therapy records, neurology records, special education records -- and the list keeps going -- to

prove the diagnosis and pertaining conditions, a diagnosis of Batten Disease, in any of the acceptable standards, would appear to much more simpler for our families.

In general, we feel that claims are made in an appropriate time frame due to the fact that we had taught the parents how to better get around the application and have a more expedited process. But if these accommodations were not made and our families were kind of going into this blindly, the process would be much slower and much more tedious for our families, for medical professionals and the Social Security Administration.

And we feel like most of the general public and, in general, the medical profession is unfamiliar with rare diseases, as you've heard, I'm sure, a lot today. Even in terms of specialists, like neurologists, the rare diseases are hardly heard about and much less seen.

With over 7,000 rare diseases, according to the National Organization for Rare Disorders, it is unlikely [sic] that medical professionals may not be as up to speed on the specifics of a disease like Batten Disease.

Batten Disease affects two to four out of every 100,000 births, so we are very rare. We have about 400 cases right now in the United States. Currently, we have, like I said, 400 children in the United States, and most doctors, including neurologists, have seen very few, if any, cases in their entire career.

So if it's hard for a doctor to provide knowledge and information on Batten Disease, imagine the reaction of a Social Security Administration worker or adjudicator trying to determine eligibility. While most parents have learned to be the medical expert for their child's rare disease, it would be helpful for large government organizations,

like the Social Security Administration, to also be somewhat knowledgeable about the rare disease, even if it just means having it listed in a book.

Providing medical records is often not a problem, but providing a large amount of information about the disease may be harder for some doctors to do. The most common problem we have is obtaining an exact diagnosis for Batten Disease. Being that Batten Disease is an inherited, genetic disorder, pinpointing the exact form and location of the gene responsible for the disease is costly and time-consuming. While all the other facts seem to be present, the confirmed DNA diagnosis can hang up the process in many areas, including social security benefits.

While collecting the medical information may not be hard, it is time-consuming, as I've stated. For children with Batten Disease, medical information can

come from many professionals in many different fields. Our children may have records from ophthalmologists, neurologists, physical, occupational and speech therapists, psychologists, classroom aides, laboratories, hospitals -- and the list goes on.

For children with rare, complicated neurological diseases, the team of specialists can also be very large. And having to collect all of this information is important, though, to determine the appropriateness of the SSI and SSDI eligibility and to give nonexperts a better overall picture of what the disease entails.

One of the most common errors, as many or all of you may know, is the decision time frame. Most families are told that their decision may take weeks or months. Most workers, when asked why it takes so long -- and this is the response we get from many of our families -- is that the worker says, "we are overloaded right now."

While the Social Security Administration is trying to determine the laundry list of symptoms and conditions that a child with Batten Disease has, it would be so much easier and faster to be able to look up the rare disease in a manual as a means of determination.

As far as the TERI programs and other programs going on right now to help expedite processes within Social Security, I feel personally, and my organization feels, that it isn't a working program because it isn't really being well-advertised. I came into this job -- I'm still very new at this, but I came in in June and knew nothing of the TERI process.

Had I known, I would have been telling my families about this all along as a way to get their Social Security applications expedited much quicker because Batten Disease is a terminal illness. It's the perfect type of program for people with rare diseases that

are terminal. However, it takes education and outreach on the part of the Social Security Administration to let rare disease organizations known about special clauses like these.

I surfed all over the Social Security Administration website multiple times and I have yet to see the TERI program, so I don't know if I'm just not looking in the right place, but it definitely is something that could be more just out there and more advertised for those of us in the profession that are trying to help families with this sort of thing.

Parents of children with rare diseases are too wrapped up in the present time with what is happening to their child right now so -- what symptoms they do display right now, that the thought of their child's life's end is not at the forefront of their mind. It may also be interesting to know whether Social Security Administration workers

and adjudicators are offering up these types of programs to parents with a child who has a rare diseases.

As stated earlier, we encourage our parents to write every symptom and condition of Batten Disease. But while the symptoms are numerous, they do not always point to a terminal illness. So while -- parents or Social Security Administration workers may not think to ask about special expedited programs for terminal illnesses.

While the program itself may work, the widespread education and advocacy about the purpose and use of the program, I feel, is lacking. Up until now, the Social Security Administration has not done too much to respond to and connect with the rare disease community. I know Mr. Astrue came and spoke at the National Organization for Rare Disease conference, and that was extremely helpful for me, somebody being new in the profession, but I think it was also very helpful for those of

us in the rare disease community, and I think we definitely felt like there was a huge outreach from the Social Security Administration, and I want to thank you for that. It seems very helpful, and it really gives us a lot of encouragement from those of us in the rare disease community.

Even though rare diseases seem like a small percentage of the country's population, there are actually 25 million people that have a rare disease, not to mention the families that are also affected. With this new attention being given to the rare disease community, I would like to encourage you to take it one step farther and begin providing the outreach and education about special Social Security Administration programs that would assist families and individuals experiencing a rare disease.

Some of the suggestions I have -- and I have a long list here; I was really brainstorming for you -- a list of rare

disorders and the basic criteria of the disease would be very helpful for adjudicators. Also, having a contact listed next to the disease like, for example, Batten Disease and the Batten Disease Support and Research Association with our 800 number could help to answer any further questions an adjudicator may have, without specifically talking with any one -- or about any one child or family.

Batten Disease is a rare disease that has many different forms, and each form can bring about symptoms at different stages and rates. This type of information is also important when determining eligibility and, thus, strengthening the argument for having a contact person on the list.

Although a list of rare diseases would be helpful, there are certain things to consider when compiling a list. In regards to Batten Disease, the term "Batten Disease" refers to the umbrella heading given to all

NCLs, or neuronal ceroid lipofuscinoses. There are currently ten NCLs that we know about, and while all are referred to as Batten Disease, each has its own distinctive name as well.

Batten Disease is not the only rare disease that would fall under this category. That is why it is important for a compiled manual to also list disease aliases. Imagine the upset and frustration for both a family and an SSA adjudicator if a disease listed on the form was not the same working as listed in the manual, yet it was the exact same disease.

Within the manual, it's also important to list all the accepted means of diagnosis for the disease. Consulting with a rare disease organization of a rare disease's top researchers will provide you with all the accepted means of diagnosis. In the case of many rare diseases, gene location for the disease has not yet been determined, and there are other ways of diagnosing a disease without

DNA confirmation.

Finally, I think I would just like to share these last few with you. I think a special financial statute should be determined for families with children with rare diseases. Because of the complications associated with the disease and the likelihood of spending more on insurance, medications, medical equipment and specialists, a family with a child with a rare disease experiences a different level of financial hardship. A family with a child with a rare disease has to make more money just to keep up with the ever-growing pile of bills.

A family may have to travel over 100 miles just to see a doctor who actually knows something about their disease. We have many families where that is the case.

In the case of Batten Disease, as in the case with most rare diseases, there is no treatment. We can only treat the symptoms as a way of treating the disease.

A child who has a multitude of different symptoms requires a multitude of medications that can be costly. The drugs that these children are on do not qualify under the new $4 prescriptions that so many pharmacies are now allowing. Medications can range upward of $300 for a month's supply, and that's only one medication.

It's also important to realize that parents of children with rare diseases -- it's very hard for them to work outside of the home -- for both parents to work outside of the home. Many parents choose to stay home and take care of their child, and while they are scrimping and saving every last penny, the Social Security Administration still determines that they make too much money.

The SSA needs to take a more realistic look at the true finances of a family who have a child with a rare disease like Batten Disease.

One of the biggest frustrations

that our families experience is the classic line, "you make too much money," which is the truth, and the SSA ends the communication. If the SSA cannot help the family, the workers need to be trained on what other avenues families can turn to for help. It seems like so many of our families see the door shut right on their faces as soon as they're told they make too much money, and they're not given any place else to go.

Finally, another suggestion would be to improve the information and communication provided by the SSA for rare diseases. In this day and age, people are using the Internet for information more and more. It would be beneficial for both the SSA and the rare disease community if a spot on the website was made available specific to benefits for people with rare diseases.

The Social Security Administration could also provide a rare disease hotline where families to call to ask questions to see

if their rare disease is listed in the manual. They would also know the Social Security Administration's preferred term for the rare disease and the specific diagnosing criteria needed to qualify for benefits.

I just want to thank you for this time for allowing us to come and speak today at the hearings. I hope my comments and suggestions have proven helpful. I'm really honored to be here. Thank you very much.

COMMISSIONER ASTRUE: Thank you very much. Before I launch into a whole bunch of questions, I should mention that we don't have statutory -- we're actually barred now from referring claimants to disease organizations. Apparently -- and this was, depending on who you believe, an intended or an unintended consequence of some amendments to the -- to the ticket to work legislation in 1999 that amended our statute.

I personally think -- would like to have that authority. It gets complicated in

terms of where you draw the line in terms of which nonprofit groups -- and we don't have an actual proposal on the table, but I have discussed that with the Hill, so just simply, so that all of you know, we actually -- I have a lot that I can probably do in my administrative discretion, and we're trying to push that as far as we can, but that's actually not one of the things right now that I have the statutory authority to do. That's a very little known fact. As a matter of fact, when I came in, I didn't realize that that law had changed in that regard. I think it's an unfortunate change.

Let me start, if I could, with Shelley. If I don't mind, I'd like to just ask you a few questions about your son's situation. And if there's anything you don't want to answer, I understand completely.

MS. BOWEN: That's okay.

COMMISSIONER ASTRUE: First, was he over 18 or under 18 when he applied for social

security benefits?

MS. BOWEN: He was over 18.

COMMISSIONER ASTRUE: He was over 18. And was he enrolled in college at that point or was he still in high school?

MS. BOWEN: He was still in high school. He was on a hospital homebound program. He had lost 60 pounds in a period of one year. He had two cardiac arrests. He had three bouts of heart failure, and his heart rate was down to 17 percent ejection fraction.

And the adjudicator spoke to me once. We had a very difficult time getting -- even getting through to her. But I understood -- I mean, it's not a common -- nobody knows Barth syndrome. I have grown so accustomed to this, that I just felt --

COMMISSIONER ASTRUE: You might want to move a little bit further aware from the mike. We're still fine-tuning the mikes here. I apologize.

MS. BOWEN: It's not uncommon for

people not to know Barth syndrome. I have just grown accustomed to spelling it, so much so that, in fact, I was speaking with Dr. Barth one time and I spelled B-a-r-t-h, and he said, "I'm familiar with it."

(Laughter.)

MS. BOWEN: So I'm just -- that's how accustomed we are to it.

Children frequently -- you know, it's just -- I'm not surprised. So I wanted to help the adjudicator in every way I possibly could.

COMMISSIONER ASTRUE: Which state do you live in?

MS. BOWEN: Florida.

COMMISSIONER ASTRUE: And how mobile was your son at that point?

MS. BOWEN: Well, he was -- you know, he was in class 4 heart failure. He was not feeling great, but he was -- he's -- you know, he's --

COMMISSIONER ASTRUE: Was he

walking or was he in a wheelchair?

MS. BOWEN: Yeah, he walks. No, he walks. As I said, these kids look deceptively healthy. If you look -- they have always been sick. My son, one of the things that he dislikes the most is when people say, "what is it like to have Barth syndrome? What is it like to have a disability?" And he says, you know, "what's it like to be health?" You know, I don't know -- that's a silly question. I mean, he's always had it.

So he functions really well. And one of the questions that we were asked -- and I wrote these questions -- and they're laden with bias. So it says, "how far can you walk without resting?" Well, we've always tried to help our son succeed. So while it takes us about an hour and a half to go around the block, where other people can go really quickly, we take one deliberate step after another with our son.

So he was answering his own

questions and, to him, he was doing very well because we have always made him able. So we had to break him down, and it was hard. It was a very hard process.

COMMISSIONER ASTRUE: It's just -- it's helpful for us to, when things are going off the tracks, to try to figure out why. You know, when we first started talking about this, I think a lot of us believed that relatively few of these cases went off track. And so rather than just have an argument, we do what I like to do -- and we actually went out and collected some data and just picked six or seven disease where we all agreed that people pretty much should be entitled to benefits with that disease or condition.

And the percentage that went off the track -- I mean, in the majority of cases we did the right thing, and we did the right thing fairly promptly. But on the other hand, you know, the percentage of cases that fell off the tracks for one reason or another were

higher than I think most of us thought that they were at the time. So it's important for us to try to figure out why that is.

And mostly it's a failure of information, and again, it's the job of the central headquarters to try to provide that information out to the field because I think most of the people in the field are trying hard, but they've got big case loads, and it just -- particularly when you get into the rare disease area, they just don't have a lot to rely on. And the general docs, too, they often don't know that much, and they go to the textbooks, and there's not much there, and that's when mistakes get made is when there's a shortage of information.

Okay. That's very helpful.

I wanted to ask you about Marfan, which I'm less familiar than a lot of the disease that we've heard from today -- could you give me a little bit more of a sense of -- you probably have the curse in that -- it's a

blessing and a curse that there are some famous people that are associated, or arguably associated, with Marfan. That may give people the wrong impression.

Can you give me a little bit more sense of the range of the disease and the scope -- the range and severity. How many people, for instance, once they become adults, roughly, just are totally incapacitated? How many people are -- you know, they may be struggling, but are actually working -- and if there are some very moderate cases -- if you can fill that in for me a little bit more, that would be helpful for me.

DR. GRIMA: For Marfan syndrome, there is a great variability within the syndrome. Most people, I would say, carry on full productive lives, but there is a small percentage -- I don't know the statistic on this, but I would say maybe between 5 to 10 percent of the population -- that ends up being more disabled because of the chronic

orthopedic issues that are happening to this older generation.

There is also a set of people that have more volatile aortic problems, that have a history of dissections in their family, that have to undergo multiple surgeries. And after having multiple surgeries, one for the aortic root, one for descending aortic aneurysms, which are even -- the surgery is much more risky -- they end up having chronic fatigue. And they have -- also with Marfan syndrome they have what's called dural ectasia, and that starts giving them pain in their back, shooting pains down their legs, so that they have chronic pain.

It doesn't happen to a high percentage. It happens to maybe 10 -- maybe 10 percent of the population.

COMMISSIONER ASTRUE: Okay. That's helpful.

Amy, I wanted to ask you just a little bit -- I know a lot of the lysosomal

storage diseases -- the severe cases and the majority of cases are ones where there is no protein being expressed by the cells at all, but there is often sort of an outlier population, 5 to 10 percent, where there is some residual ability to produce protein, but it's limited.

Is that the same way it is in Batten's?

MS. KIRK: Yes, it's exactly the same.

COMMISSIONER ASTRUE: And what is the 5 -- the 5 to 10 percent who are least severely affected, maybe if you could sketch out what it -- can you make any generalizations about, typically, what their lives look like?

MS. KIRK: For many of them, it may be just be a delay of the onset of symptoms, so their body is producing enough of the protein where their seizure activity might not quite be as severe as somebody that has -- is

making less of the protein.

But, generally, the symptoms are -- I mean, it's not like a symptom isn't ever going to show up. It will definitely show up.

It is very rare that it's happening to somebody -- I mean, you definitely know that there's something wrong. It's not like they're going on throughout life and you're not seeing anything is wrong with them.

COMMISSIONER ASTRUE: Do you know anything -- I mean, if you have a study, that's great, but do you have a gut sense for what life expectancy is for --

MS. KIRK: It's still very low. It's still -- I mean, they're still going to pass away, unfortunately, but --

COMMISSIONER ASTRUE: Are you talking teens or 20s and 30s?

MS. KIRK: If they're lucky, 20s or early 30s. I mean, and that is very, very -- if -- their body has to be producing an unusual amount of the protein. But very

rarely are they out of their -- early 30s is long, and that's for just the juvenile-onset. But if it's infantile, or late infantile, it could be late teens, mid-teens, early 20s, somewhere in there, but very few past 30, early 30s.

COMMISSIONER ASTRUE: Okay. Thank you.

MS. KIRK: You're welcome.

COMMISSIONER ASTRUE: Steve?

DR. GROFT: My apologies if these questions were answered earlier. Yesterday we heard from Ron Bartek from the ataxia group, and he talked about how he took a rating scale from -- multiple sclerosis, I think, was adaptable to that. And two of you had been successful, I guess, in getting the determination made -- or your organization having the determination of disability made.

Do you put up a template -- or how do you make the information available to your members that this is a good way to go, to work

with the Social Security Administration? Do you do anything like this, or provide advice or guidance?

MS. KIRK: They hired me. That's how they --

(Laughter.)

MS. KIRK: I mean, before I was hired, it was the executive director doing all of this, and it was just a lot of -- he doesn't -- I mean, he only had experience himself from being the father of a child with Batten Disease and being executive director for as long as he had.

He was really -- I mean, it's just really trial and error for us and learning what families can do that's helped and what families aren't doing, and so we kind of, after a long amount of time, we just kind of came up with, you know, it helps a lot more if you individually list out your symptoms of your child with Batten Disease.

Instead of putting Batten Disease,

because they're not going to know what that is, if you put blindness, if you put loss of speech, seizure disorder, all that good stuff, they're going to be more likely to say, "wow, this person really does have a major disability" instead of just putting the one-word title, Batten Disease.

So it was very much trial and error coming about, so...

DR. GROFT: Okay. Thank you.

Shelley?

MS. BOWEN: We -- Steve, we actually -- our children -- we were just established in 2000, so we're just now beginning to see these young men come around into their second decade of life.

The other thing that I think is important to note is that we are primarily a virtual group, and so we have high-functioning families that are in our midst. Most of them have -- they have access to a computer. They have one in their home. They plan for their

children to go to college. These are not people that would necessarily even think to ask for social security for their children.

It was interesting -- I did a survey before I left, and 67 percent of the families polled never even considered asking for assistance even though one of the parents had to quit work, the child was frequently in the hospital, they had to claim bankruptcy -- you know, it's absolutely amazing that they don't even think about -- they think there's other people out there that are more needy than I am.

So, obviously, this is something we, as a group, have to internally address as well.

DR. GROFT: I mean, you're absolutely correct. I think one of the problems and one of the concerns we have dealing with rare diseases is that we know we have access to those who have ready access to the Internet. But there is a significant part

of the population that just does not have ready access to it, and, you know, we just have difficulty reaching the entire population here in the United States. So we're suffering the same problems, too.

Another question, if I can. You know, we're looking at maybe 6, 7,000 different disorders, and we certainly can't attack -- when I say "we," working with the Social Security Administration -- any suggestions on how you pick 300, 500 diseases, 100 diseases? Any thoughts at all?

I'm sure they -- the Social Security Administration has thought about this. But as a patient group -- I mean, I think everyone wants to be classified and be on the list -- and Diane Dorman yesterday talked about low-hanging fruit, although I don't think she used that term, but something like that where you can pick off various diseases that have very, very, you know, serious disabilities -- but any thoughts or

any other way -- how do you create a list?

MS. BOWEN: I was thinking about this earlier because I was listening -- you know, you guys are sitting in the hot seat. I thought I was sitting in the hot seat, but we're actually saying, you know what would be great: Here, we'll just make a list of the diseases.

But you're right -- I mean, coming up with a mutation doesn't necessarily mean that it's a disease-causing mutation or that it's going to be -- immediately it's going to be a disease-causing mutation.

And I was sitting back there -- you know, I would not -- I just happen to be one of the lucky ones that fell off the track, you know, but at least I'm able to get up and say, "this is what's happened," and then consider the other side of it as to what we can do.

But while you can't talk to disease groups, we can help provide -- look at the symptoms -- and there's got to be some common

symptoms of rare diseases, and I've talked to Ron Bartek today, and then there's Duchenne muscular dystrophy -- and a number of these diseases have common issues, such as cardiomyopathies. And so coming up with, you know, when is it debilitating? What does it look like? What can -- you know, what can we help to do in terms of better describing the cluster of diseases?

And the other thing that was really interesting on the website is I didn't see anything about mitochondrial diseases listed on the disabilities on the website, which are pretty debilitating disorders.

So that was the suggestion -- that would be good. But I would be willing to do whatever I could to help.

ACTING DEPUTY COMMISSIONER RUST: When we adjudicate cases, we are -- there is always a gray area -- I mean, this is a problem we have. There's -- very few people present themselves that are clearly not

disabled -- but a few do -- and many that are clearly disabled, and then you get the center area.

For each of the three of you, are there beginning to be evolved objective standards or scales or measures that the physicians use or the researchers use to begin to identify the degree of severity of the condition?

Shelley, can you start?

MS. BOWEN: We just established a medical database and biorepository to develop longitudinal data. And we enlisted a university that has a biostatistician that can help rule out the bias there.

You know, it's really -- it's very subjective, and we're hoping that we will be able to come up with better standards by collecting longitudinal data. I think this is a good question. I think it's -- it's one of those future things that we need to do, but it is a -- it's a struggle.

DR. GRIMA: For Marfan syndrome, we do not have anything really like that. There is a lack of medical evidence showing the functional disabilities for patients with Marfan syndrome. A paper came out recently, within the last two years, beginning to look at that, at the pain associated with the disease, mainly looking at orthopedic issues.

And -- because this is -- it's becoming a new entity. It's becoming more apparent in this group because they're older. Usually, patients with Marfan syndrome died at a young age. We didn't have an older population, and so now we're getting into a new set of debilitating issues, their chronic issues, their pain and their fatigue, and it's very hard to actually put a number on that.

So we're beginning to have scientists look at that, but there is really nothing that is in, you know, medical evidence for that at this time.

MS. KIRK: As far as Batten

Disease, I mean, we know the general progression of the disease, so we know what each stage is going to kind of entail, but our children are so different as to who kind of hits what stage and when they hit it -- not all children will hit those stages, so it's very hard to get a real clear-cut cookie cutter kind of picture.

And it's very subjective as to what you would say is severe versus not -- you know, functional versus very severe. I would say that a feeding tube would obviously be very severe, but at the same point, a child that is having mobility problems and blindness with, you know, multiple seizure activity every day would also be severe. So, unfortunately, I don't think we have really that very clear-cut data. It's still very subjective, so...

JUDGE CRISTAUDO: I have several questions. Again, Ms. Bowen, if you don't want to answer this question, that's fine, but

did I understand -- was your son ever approved?

MS. BOWEN: No, he wasn't. In fact, I consider myself as one of those people that didn't really know what to do next.

I didn't -- the last statement on the denial said that if he were -- if your condition worsens, you can always reapply.

So at that point, he was in heart failure. He had lost 60 pounds. He was pretty bad. You know, there was nowhere but down to go. So we had -- we didn't -- I let it go, and my sister-in-law, who is a social worker, chastised me for not -- she said, "well, you don't know the system." And I said, "I shouldn't have to know the system." I mean, it shouldn't be a system. I think -- it's tough. You know, it was really hard for me.

So -- and I'm extremely optimistic and altruistic, so I believe in the greater good of the government and the world, and

everybody is good and glorious. So I just thought, well, they must have thought that he didn't need it, and so --

JUDGE CRISTAUDO: Did you go to a hearing?

MS. BOWEN: I reapplied, and we'll see what happens. That was a year and a half ago. But he has nothing right now, and I put it on -- I tried to get it on the fast track. We'll see what happens. That was two weeks ago.

JUDGE CRISTAUDO: Did you go to a hearing the first time?

MS. BOWEN: I never had a hearing.

JUDGE CRISTAUDO: There were some comments about the TERI process, and certainly, as you suggest, we do have a process where, if someone does have an illness that -- that -- they are dying, certainly, or if there's some other situation that's really very dire, they're in foreclosure and so on, we do have a process where we will try to give

priority, certainly, to those cases.

And I think the suggestion was is that we need to be advertising that more -- is that what you're suggesting? I mean, I guess -- you know, there's a little bit of an assumption that if someone's condition gets to that level or if they're in foreclosure, that they are letting us know. But I think you're suggesting, in fact, people -- it could be happening to people and they're not letting us know. We never become aware, in fact, that their condition is like that.

MS. KIRK: Well, with our families -- with -- I mean, as I kind of -- when families go in and they list -- as we've taught them to list every single symptom and condition of their disease, a lot of times terminal illness doesn't come to mind, or parents just -- they don't know. They don't know that, because their child has a terminal illness, they get a speedy process.

And I don't think they're getting

asked, "is this a terminal illness" during their process because, from the parents that I've experienced in my unfortunately short time there, I've never heard of a family who was in the TERI process when applying for social security. And I didn't even never knew about it until I read the witness questions. So -- and I've been doing this since June. So this is new to me as well.

Now that I know, I am obviously going to advocate for my families -- that's the first thing they're going to ask for. And I called my local social security office, just trying to probe a little bit and see what I could get out of them. And, you know, I asked, "is it a separate form?" Because I didn't even know. "Is it a separate form from the regular -- let's say they're applying for SSI. Is it a separate form?"

And, you know, the first office I called, they didn't really want to tell me too much, and they just kind of said, "well, you

come in and we'll just take care of it."

Then the second office I called was much more -- they were much more -- giving me more answers. You know, we'll do it -- you know, sit down with them -- they just gave me a lot better answer. But, I mean, it sounds to me like -- and the way that I feel about it is that I just -- I don't think that the rare disease community knows that this program is out there, because I know my families aren't using it, so...

JUDGE CRISTAUDO: The last question: If I understood you correctly, obviously, there are a number of medical sources that are not aware of the three conditions, certainly, that all three of you have talked about here today. Are we having situations where they are reporting essentially symptoms and so on, but they're never making a diagnosis of Marfan or Barth and so on?

Is that happening where no

diagnosis is made; they know there is something going on, but they don't know what the -- what the actual disease is, or what the impairment is? Is that what you're suggesting?

DR. GRIMA: For Marfan syndrome, there's a definite diagnosis. I think, when it comes to Marfan system, what the problem is is that they don't understand how it can be functionally disabling. And I think that's where the question is for Marfan syndrome.

JUDGE CRISTAUDO: How about the other two? Because it sounded like what you were getting at is that -- these are obviously -- in one I think you said 400 cases in the whole country, and the other perhaps not very many. There may be situations where, in fact, people have an illness that is leading to death, or clearly disabling. They have all the symptoms, but they're not able to make the diagnosis. I'm trying to figure out what we should be -- any recommendation you

may have in terms of what we should be doing with that situation. I mean, how do we deal with that situation?

MS. BOWEN: I can give you the statistics for diagnosis for Barth syndrome. It takes an average of three to five years to diagnose it, even today. Typically, it's first classified as not Duchenne's because you have these kids that are weak, like Duchenne's children.

But it's a very tough disease to diagnose because, as I said, these kids are very -- they are articulate, they're cute, they look deceptively healthy, and then they just get very sick and go downhill.

So typically what happens -- and there is also a high incidence of no -- a de novo mutation and the mothers either being carriers or the boys are carrying. So you won't see a family history, so oftentimes it will be the death of one son, and then another son being warned and developing the symptoms

before you'll see the disorder.

And it is extraordinarily difficult to take care of early in life. The children have a lot of eating problems, so it's not uncommon for a child to die within the first couple of years, and a parent would never even know what's going on.

MS. KIRK: Similarly, with Batten Disease, the average process -- I mean, it can take three, five -- sometimes the children still don't know and they've had the disorder for ten years because it is so rare that the first sign is usually vision loss, so they go to an ophthalmologist or an optometrist -- and they have retinitis pigmentosa, or some other ophthalmological disease.

And then they have seizures -- then they start having seizures. "Well, you're just unfortunate; you have a kid that has epilepsy and blindness." And it takes a long time before, finally, people start picking up on the fact that there's something else wrong

with these children.

And because there are so many different forms, while a child that starts with these symptoms at age ten looks like they have a juvenile presentation, and you test them for juvenile with DNA, because that's really -- I mean, that's a confirming diagnosis -- then you don't have juvenile. So then you're back to square one with searching as to which type of Batten Disease that they actually have.

And we have a lot of families out there still that don't have diagnoses, but it is clearly Batten Disease, as the brain atrophy, the occlusion bodies on their skin biopsies and things like that -- all the other signs are pointing to Batten Disease; we just don't have a clear-cut DNA confirmation diagnosis. But all the other things point to it.

JUDGE CRISTAUDO: Of course, some misdiagnosis -- do we have many cases where

there is, in fact, no diagnosis made?

MS. KIRK: Ever?

JUDGE CRISTAUDO: Or are they just simply misdiagnosed?

MS. KIRK: I'm not sure. I can't answer you that. I know we have families out there who are still trying to find a diagnosis. And a lot of times if it -- it looks like Batten Disease, but it might not be, so we might say that it isn't. It can be very confusing. It can paint a different picture, and it might actually be something else and look like one thing.

So I'm sure there is, but I unfortunately am not really positive on that.

MS. BOWEN: I don't think we would know either. I think they would contact us because they either suspect there is a diagnosis or they have been diagnosed with it. And our sense is that the doctor sees that there's so many -- the entire gene and intron and exon are not sequenced. So it could

easily be that there's something going on in the MRNA, or something going on in the protein products. So if the kid has the symptoms, he has Barth syndrome.

COMMISSIONER ASTRUE: Frank actually asked -- I mean, that's a really interesting question that I hadn't really thought terribly hard about before now, because it's -- whenever you have a system as big and complicated as ours, you can't help but think in the categories of the system. I mean, it just becomes engrained.

And I think it is actually much more common certainly than a lot of people in the system appreciate to have people with extremely serious problems where they never do have a diagnosis.

I've got a very close friend who's got two children with the same syndrome. And they've been out to NIH a million times. They've been at every major medical center, and they can't get a diagnosis except that

it's -- well, it's sort of like autism in certain ways, but it's clearly not autism. And they've been looking for a long time.

And I suppose that that's one of the things that we probably ought to take a look at because, you know, if you can't put a label on the disease, you shouldn't really be at a disadvantage in the process. And I've never even asked the question whether, you know, when someone comes in and they don't even have a clear diagnosis, sort of how the system handles it.

I mean, theoretically, I think the way it's supposed to work, they shouldn't be at a disadvantage as they go through. But there may be some unintended common misconceptions and things like that, so that's probably actually something we should take a look at and just make sure that, you know, there's nothing -- I think that's a very good thought.

DR. GROFT: If I could just make a

comment. During the last National Commission on Orphan Diseases, as I mentioned yesterday, we did a small survey of patients with known diagnoses of rare diseases, and at that point approximately 15 percent of the patients, it took longer than five years to get a diagnosis. That meant a lot of doctor visits, a lot of multiple hospitals, and moving up and down within the system. And even then, I think we had 31 percent took between one and five years to get a diagnosis. So we have a lot of patients visiting many, many physicians.

And NORD did a similar survey in the early 2000s here, and I think they repeated those numbers. So it is a major problem, getting a diagnosis. And, certainly, undiagnosed diseases, or diseases of unknown origin, whatever you want to call them, it is a significant problem for patients and for the physician. I mean, everyone is very frustrated because you just don't know what to

put on, and I think, regardless of the disease, if the patients and their families just have a name, they feel -- at least they can start to look for information. Maybe they can find a physician who has that knowledge.

So it's a tremendous task, sometimes, to get that diagnosis.

MS. BOWEN: But, Steve, it's also -- don't you also think that we're -- we're in an age where we're more refined, so no longer is it just heart failure; it's heart failure because of 141 possible causes, and it could be a genetic cause or it's years of abnormal metabolism that may happen. It's -- it is -- you know heart failure is heart failure, but it is important to know what it is, and it may be that we're not doing such a bad job -- we're actually doing a better job and suffering because of that.

DR. GROFT: Oh, definitely so. I think the awareness of rare diseases has increased tremendously, at least in the last

25 years that I'm aware of, and it's still growing, so it's good to see, but yet every time you feel you have a success, you have five other diseases that you just don't know anything about. So that's what keeps us going.

COMMISSIONER ASTRUE: Steve, I have sort of an off-the-wall question that actually I just want to ask you, although if any of you know the answer --

DR. GROFT: They may know.

COMMISSIONER ASTRUE: We all have human limitations. It's unreasonable to expect, you know, most physicians to have a handle on all the symptoms of all 7,000 rare diseases.

Is there any kind of computer program, using artificial intelligence techniques, where, if a doctor is having trouble diagnosing a patient, he can plug in the symptoms, and then there's at least a list of what -- the possible diagnoses?

DR. GROFT: Yes. There have been several, I think, systems that have been tried, but none of them have been found to be optimal. There's too many confounding symptoms that come into the picture: Laboratory tests, imaging results -- and all of a sudden you think, well, it could be this or that -- and you just don't have that differently diagnosis that pops out at you many times.

It's something we really would like to see, and I think perhaps over time, as we start to do some more of the genome-wide association studies where we start looking at genotype/phenotype correlations -- we're going to be able to get some more of the natural history, and that -- there's correlations that will make some sense and that we can draw conclusions easier, but that's still several years out. But it's starting.

ACTING DEPUTY COMMISSIONER RUST: Ms. Bowen, you mentioned that many of the

parents never applied for benefits.

MS. BOWEN: Right, 67 percent.

ACTING DEPUTY COMMISSIONER RUST: 67 percent never do it?

MS. BOWEN: Right.

ACTING DEPUTY COMMISSIONER RUST: You also made another comment, sort of a side comment, that really intrigued me, and that is part of it is it's because they don't see their children as being disabled, or they haven't thought of their children as being disabled.

MS. BOWEN: Right. They -- I think it's -- there is -- again, these children look -- they look good. They look good. And I can remember taking my son into the emergency room, being in heart failure, and the next day we were essentially told that he was going to die, and being told there's nothing wrong with him, you know, I was just overreacting.

It's very deceptive. They look

fine. And then -- this is all they know. This is all these families know. And so they -- they're high-functioning. They have computers in their homes. One of the parents are able to stay home with them. They're able to ask questions.

They are -- these are the people who come to us. These are the ones that we know about. But the ones that I -- you know, and they're struggling. They're on the juxtaposition of the families that are really in need that you typically think of with social security and disability -- you think of these indigent children and kids that are at need that don't have insurance, and you don't think, well, just because I have to claim bankruptcy and my wife has got to quit or we've got to sell our house -- at least we can sell our house.

I mean, this is what I'm up against with these families. You know, they haven't -- they just don't think -- they think

that you have to be 65 years old. That's the other misperception: "I thought it was for only people who are 65 years old or older."

ACTING DEPUTY COMMISSIONER RUST: Is this a problem with the other two, here, Amy and -- do you hear the same sorts of comments applying for benefits?

MS. KIRK: They get very frustrated because they know they make too much money, and so they don't even -- I mean, I think a lot of our families just know that they make too much money, so they're not going to apply. But I also think then that they don't -- they kind of leave it at that, and they forget about the Social Security Administration. And I think a lot of them forget, after their child turns 18, that their child can go and apply for social security disability benefits.

So I think -- we have a lot of families, too, where it's upper/middle class, or middle class, and they just simply -- or they just make too much money, and so they

leave it at that, too; they don't go back and try and reapply when their financial situation changes.

Yeah, I agree with her completely. We've experienced a lot of the same things, too.

DR. GRIMA: I think, for the Marfan population, it's not really like that because usually they are more adults that are applying for disability -- they were working and they were -- you know, having a steady job, but then just as their symptoms get more severe and they find out that they can't hold down a job anymore, then they look to see what else that they can do to get insurance and to get, you know, other coverage.

COMMISSIONER ASTRUE: Okay. I would like to thank the panelists very much. This was very helpful.

We're going to move now into taking comment from the public. This is probably an opportune time for me to thank Diane Dorman

and the National Organization of Rare Disorders for getting the word out on this. I know we've had a lot of interest, and some people that want to speak today, and I think we've probably promised the first couple of slots to a couple of specific disease associations, but be patient if you're waiting out there. We'll get to everybody.

So I think -- if I remember correctly, we've got representatives from Tourette syndrome here that would like to speak.

MS. BAKER: Thank you, Commissioner Astrue, and fellow affiliate members, for the opportunity to present our view concerning the eligibility screening process for Compassionate Allowance for Tourette syndrome. My name is Nancy Thomas Baker, and I'm here on behalf of the Tourette Syndrome Association and its members. I have a 12-year-old daughter with Tourette syndrome.

As you may know, the TSA is the

only national, voluntary nonprofit membership organization dedicated to identifying the cause, finding the cure, and improving the quality of life for individuals with TS.

Tourette syndrome is an inherited neurological disorder characterized by involuntary movements and sounds that are known as tics. The disorder is often accompanied by attention deficit hyperactivity disorder and/or obsessive-compulsive disorder. There is no known cure for TS.

We commend SSA's initiative to provide a quick and deliberate process for determining eligibility for public services by SSA. However, we strongly urge the consideration of additional provisions that would recognize and cover serious disabilities incurred by patients with severe TS. TS is a neurological disorder, causing significant health impairments among the more severely affected.

In our view, clearly, this

condition should be recognized officially as a disability under Compassionate Allowance. Under current circumstances, we believe that people with TS do not receive a timely or deliberate -- or deliberative eligibility decision due to a lack of understanding of the disorder and its effects on some individuals.

The TSA has informally surveyed key physicians who are responsible for TS clinics, and stakeholders in several regions of the country. This survey indicates that even for those who eventually did receive approval for SSA benefits and services, the process was unacceptably lengthy, with patients having to pursue an endless appeals process.

For those with disabling TS, motor tics, as well as inappropriate prominent vocal symptoms, the quality of life can only be described as nonexistent and often intolerable. Some of most disabling factors that make it nearly impossible to cope with TS symptoms include self-mutilating behaviors,

social isolation and employment discrimination due to public stigmatization.

These very troubling symptoms are just a few examples of the most disabling outcomes that result from having this complex disorder. Therefore, we recommend that a comprehensive document be developed that would provide specific guidelines for a determination of patient eligibility. Furthermore, we suggest that SSA determine disability based on a case-by-case basis.

These would include, but would not be limited to, diagnosis, symptom severity and impairment, as well as quality of life impact. Because, unfortunately, we do not fall under most of SSA's expedited processes of claims, we cannot answer some of the specific questions about how these processes are working with individuals with TS.

However, we would like to reiterate that increased SSA understanding about Tourette syndrome, with inclusion of accurate

medical information, will result in fewer unwarranted denials and appeals. This improved knowledge will improve critical support for people with TS.

The TSA sponsors a multidisciplinary national medical advisory board whose members are available to guide and advise the SSA on all matters regarding TS patients. We urge the SSA to take advantage of the expertise of the Tourette Syndrome Association's medical advisory board by consulting on the objective, medically evidentiary requirements for TS eligibility.

As a mother of a 12-year-old with Tourette syndrome and a board member of the National Tourette Syndrome Association, I would like to request that the SSA consider TSA's views and make every effort to serve our most vulnerable citizens with Tourette syndrome. We recommend that SSA expand the list of impairments to include Tourette syndrome.

Thank you so much.

COMMISSIONER ASTRUE: Thank you. I should mention that we have -- we're in the process of doing overhauls of a number of our body systems for the listings. We've got 14 areas in our current regulations.

Neurology is not next in the line, but it's right up there near the top of the queue. So it is -- your comments are very timely because we'll be revising our rules in the neurology area generally over the coming months. And so getting any detailed comment in on what you would like to see should be very helpful.

And in particular, as I said at the outset, we have our ordinary procedures and listings, and we don't want to overlook that. We want to make sure that those are right, too.

To the extent that there are going to be extremely severe cases that might qualify for one of our expedited review

systems, to the extent that you can give us help on how to sort out the more severe patients from the less severe patients, that's extremely useful information to us as well.

MS. BAKER: Very good. There is also a video that HBO -- a documentary that they did on Tourette syndrome, and in that -- we can provide you with the copy of that, and also the medical advisory board -- it's very clear, you know, the severe cases -- my child's is moderate to severe, and over time it's improving. But the severe cases, it's obvious just to the eye. I mean, the tics are so pronounced --

COMMISSIONER ASTRUE: Well, I don't mean to be difficult, but one of the things you have to -- you have to put yourself in my shoes for a moment. We've got thousands of people that work for state agencies around the country that make these decisions for us.

And we really -- again, no disrespect to them. I think -- you know,

they're hard-working, wonderful people, almost to a person. But it's unreasonable to expect that they're well educated on every disease and condition. And what we have to do -- what our job is in Baltimore is we've got to break it down as simply and clearly as possible.

And I know -- it's got to be frustrating for patient advocates because it's so obvious to all of you -- and it may even be obvious to us up here. But what we've got to do -- our job is we've got to find a way to make it obvious to people that don't know anything about Tourette syndrome or Batten Disease or something like that -- so to the extent that you can help us put it into words so we can tell people out in the States and we can tell our administrative law judges how they ought to be looking at it -- anything you can do in that regard is very helpful to us.

MS. BAKER: Absolutely. Thank you.

COMMISSIONER ASTRUE: Anybody else?

Diane, I've got such an overload of

information, I know I promised the next slot, but now I've forgotten -- oh, yes.

MS. LEWIS: Thank you very much for the opportunity. I'm Rosalie Lewis from the Dystonia Medical Research Foundation, and along with all the other advocates who have spoken today, as -- I would like to thank you for the opportunity for all of us. And I have to say, I have been so educated by the advocates today who have presented, it's been a marvelous time.

In any case, I have been involved with the Dystonia Medical Research Foundation for the last 30 years when the first of my four sons were diagnosed with this neurological movement disease, three who have the disease actively, and the fourth is an asymptomatic carrier of the disease.

I represent the foundation as an immediate past president and as the current vice president of public policy, and also as the chair of the Dystonia Advocacy Coalition,

which represents five other dystonia groups within the United States.

The Dystonia Foundation was founded, just like many other advocacy groups, with the intent to raise awareness and educate medical and lay community to do support services for our community, as well as to fund medical research. And we have been very successful in undercovering -- uncovering, rather, a lot of new science, but in the process, as you can well imagine, that leads to the next question: More and more of the science is -- it's not understood. And, in fact, when I speak about dystonia, people look at me and say, "is that a country in Eastern Europe?" So we do have a lot of work to do every single day of our lives.

So what is dystonia? Well, unlike several of the diseases that you've heard of already, dystonia represents about 300,000 people -- and that's a conservative estimate -- in the United States alone. There

are so many different forms of dystonia, that, in a composite, we say they can be as much as a half a million.

However, the focal dystonias represent a smaller population -- and I'll get into the different forms, to give just a little bit explanation about why it's so difficult for the Social Security Administration, and others, to say, "oh, this is dystonia, and this is a severe illness."

So dystonia is a neurological movement disorder that affects the muscles of the body, causing contractions, spasms, pain, chronic issues, and it's progressive. So there is a form of generalized dystonia that my children have -- it's a genetic autosomal dominant form -- that starts at around the age of seven. A child is born looking perfectly normal. When they get to a developmental stage in their lives, all of a sudden they can't walk, they can't use their hands, their voice might be affected as well as possibly,

but not very likely, torticollis where their head is pulled to the side or the back.

From that point at age seven on through their adult years, the disease progresses. There is no cure for dystonia. There are medications, but the medication side effects oftentimes make the disease almost worse than it is.

The focal dystonia start more in a person's 40s to 50s, just when they are the most productive in their careers. It can be so disabling, that a person who develops blepharospasm, which is excessive blinking of the eyelid, may become functionally blind. They can't see well enough to walk out on the street by themselves, cross the street, read a book -- certainly not to drive.

People who have oromandibular dystonia have it so that their jaw is clenched shut. They cannot speak, nor can they eat. Spasmodic dystonia is a focal dystonia of the vocal cords where production of sound becomes

constrained -- or it sounds like this -- or it's very, very whispery. One of my sons developed that in addition to his generalized dystonia, and even botulinum toxin injections into his vocal cords which were not very helpful.

Focal dystonia of the arm prevents the person from writing. Their hands become contracted like this, so if they're trying to hold a job or they're trying write, trying to use a computer, they can no longer do that. It's an involuntarily contraction, usually as a result of trying to do a job.

When a person in dystonia sleeps, the muscles are relaxed, and everything is calm. As soon as they're up, the spasm starts all over again.

Another form of focal dystonia -- and you've heard this in musicians. Musicians have developed dystonia, and that makes them totally unable to continue their career, whether it be in the muscles around their lips

so they no longer can play a wind instrument, or they no longer can finger a guitar or any other stringed instrument.

Or a conductor of orchestras. Normally they can use a baton and keep it going.

Focal dystonia has also affected Leon Fleisher, the pianist. He can no longer play the piano until recently when the Botox shots started to work. In fact, he just won an award at the Kennedy Center the other night.

So there are many genetic forms of dystonia and then there are forms of dystonia that come as a result -- secondary to injury, traumatic brain injury, a limb injury or an injury -- or subsequent to disease -- medications for diseases not related to dystonia but cause dystonia.

There are people with Parkinson's disease who have dystonia, as well as people who start with dystonia that develop

Parkinsonian-like symptoms.

So you can see the range of the disease. It can not be terribly severe to so severe that a person is twisted up into a pretzel -- and that's one of the descriptive terms, pretzel-like. And if you saw a person -- specifically a young child -- with severe dystonia, they can't dress themselves, eat, certainly cannot walk. It's really most scary.

I heard you ask some other people about a rating scale. Yes, in dystonia we do have a rating scale. It is a very subjective scale, as you can well imagine. My son who was going -- and did have deep brain stimulation surgery to correct the symptoms of generalized dystonia had the rating scale performed before and post surgery, and the results were -- are remarkable. It was a tremendous success for him, thank goodness.

Other people, though, had the rating scale done in order to qualify for

benefits within their health insurance, and it might be very helpful within the social security application process.

I have my notes, but to tell you the truth, I guess the easiest thing is just to tell you from my heart. As I have been involved with the Foundation for 30 years since my children were diagnosed, I have seen adults with their focal dystonias have their lives destroyed. When it hits, it hits almost overnight. Nobody understands what's going on. They go to their doctor. They say, "oh, it's a psychological issue. Stop drinking, or drink."

Nobody initially knows what to do.

But when you see a patient, with a friend, walk with their head stuck to their shoulder because they can't pull the muscles straight back, focusing this way, you can't walk; you'll bump into everything.

Or a person whose eyes are shut tight. They no longer can even take care of

their children. It's a terribly disabling, severe disease.

And I appreciate the difficulty from your end to look at a person that says, quote, a dystonia without fully understanding the full scope of the disease.

And I appreciate what you just said, the woman from the Tourette's Association. And the Dystonia Medical Research Foundation would be happy to supply you with information that might be of benefit as you look through the neurological diseases.

We also have a very fine medical advisory group worldwide who can help with any responses to scientific questions, clinical diagnosis questions -- we have a dystonia study group who are trying new medications and therapies. We're seasoned, and yet we're very new.

I really thank you for the opportunity to discuss this because there are 30,000 people just within the Foundation who

are really eager to have your help.

COMMISSIONER ASTRUE: Thank you very much. Certainly the scale would be helpful to us, any studies associated with development of the scale and the validation of the scale is very important.

And if you've got a couple top-notch docs who, you know, would be willing to talk to us about the very -- this is obviously one of the ones that's very complex from our point of view to try to get a handle on, but, you know, forwarding to us some documents that we could ask questions to once we look at this again, I think that would be very helpful to us as well. So I thank you.

Gentlemen, anything else?

JUDGE CRISTAUDO: I would like to ask the last two witnesses if you can just comment very, very briefly on -- for the individuals who have the conditions that you have described, what's been the experience in dealing with our process? Are people who are

applying with these conditions, are they being approved pretty quickly, or is it not happening that way?

MS. LEWIS: So I asked, in this survey, on my own website, and it's anywhere from six months to two years.

Children get approved much more quickly. Adults don't. And sometimes -- I think it's about 80 percent do get approved finally if they go before the judge. If they don't, it gets rejected, rejected, rejected.

JUDGE CRISTAUDO: Thank you.

MS. BAKER: We are not sure overall -- it varies case to case -- how long it takes to get approved. And -- because we're getting the information not from the patients themselves, but from medical professionals who are dealing with the patients. So the TSA is now gathering information on a case-by-case basis.

JUDGE CRISTAUDO: Thank you.

COMMISSIONER ASTRUE: My staff has

made a very important request, which -- they've asked me to define the word "us" when I said, "get in touch with us." So you'll find that -- although I have the best of intentions, I get such a huge volume of mail, that sending it to me directly, while that may seem logical, isn't probably in your best interest. So we do have, in the materials, the email address for comments, and that generally -- for everything that we're discussing today, generally is the best place to do it.

And if I remember this correctly -- and this is from memory -- but it's compassionate.allowances@ssa.gov.

And that really is the best place to direct any comments that you have.

Those are the two that I knew were here, but there may be others here. Is there anyone else in the audience that wants to add to what we've heard already today? Going once.

MR. YUKAN: Hi. I'd like to thank you. My name is John Yukan [phonetic]. I have a child with juvenile Batten Disease, ten years old.

I was blessed because we got a diagnosis within 18 months. Most of these kids don't get -- with juvenile Batten Disease don't get it diagnosed until the ages of 15, 16, when seizures begin. And it's just such a frustrating feeling because every social worker we've talked to says, "you qualify; you qualify." But every time we come back, it's income.

And it's -- it's not even an answer of where to go from here. It's just, "okay, you don't qualify. Thank you very much."

And it's just such a frustrating feeling because you hear from all the workers saying you meet everything, but you don't.

And we've -- you know, we've even started looking at the autistic waiver. We've got a social worker that's looking at that

because a lot of the children with Batten Disease have a lot of the autistic symptoms, but some people don't want to fill out the paperwork because it doesn't relate to Batten Disease. Thank you.

COMMISSIONER ASTRUE: Thank you.

Anybody else.

MS. BOWEN: I have a question for you. You get to ask us questions; now I have a question for you.

COMMISSIONER ASTRUE: Hey, wait a minute. I make the rules here. No, go ahead. You ask whatever you want.

MS. BOWEN: You know, you are in Washington, D.C., and this is -- you guys get a lot of information, and I just want to commend you for being so receptive.

And I just did an organizational evaluation for the few children that I take care of, and it was just a cacophony of noise. I was ready to go nuts. But sometimes it's just good to just get away and put people

together in a think tank about what the issues happen to be.

And it occurs to me that this is not just about social security, this is not just about rare diseases. This is about new survivors, better medicine, new issues that are occurring. And we are in an advent of the technology now where we can create health records, electronic health records that can go with us, with our children wherever we go.

But somehow there's got to be a way where you guys -- I will cook dinner. You can come to Florida. We'll do a cook-out, just chill and fish and talk.

But does that ever happen? You know, besides watching C-Span and then, when it pans out and there's nobody in the audience -- that's my extent of, you know, being able to really see discussions happening here. What happens?

COMMISSIONER ASTRUE: Well, it's real hard -- you know, we -- particularly when

you get an organization as large as this.

It's very hard to deal with things except through a hierarchy and process and things like that. And we need a lot of that to do our job. We would break down and not be able to function. You can't manage 60,000 people in a highly fluid way.

But if I understand what you're saying properly, I think I agree completely, and it should at least be reassuring that I think it does happen. So when I recuperate from these two days -- I start off tomorrow, and we have actually taken most of the senior staff off-site to develop a new strategic plan for the agency, because we have a lot of challenges.

We have, because of demographics, our workloads are going up. Because of new Congressional mandates, our workloads are going up. And we've been below the President's budget for 12 consecutive years. And the only way out of that box is to think

about some dramatically new ways of doing our business through technology, through different procedures, through involving other entities in different ways than what we've done before.

So, in fact, a lot of the senior staff is going off-site tomorrow -- that's the kickoff for a new strategic plan for the agency.

We've also been trying to do this -- and I think this has gone pretty well so far; we're not there yet, but the woman sitting directly behind you is cochair of a group that's working with the 50 states and four other jurisdictions that run the disability determination systems for us -- to work on something that may seem arcane compared to what we've been talking about today for most of you, but it's critically important to what we do and how we do it, which is to see if we can come up with a unified IT system.

Right now, the states have

COBOL-based -- you know, which is state-of-the-art 1970 -- code, and it's making it increasingly difficult for us to adapt and plug in new technologies. And each of the states has a different system. There's some commonalities. There's several contractors that did the -- but basically we're running 54 different IT systems, and that makes it harder to adapt to change. That makes it harder to tell from my position -- or probably more important, Dave Rust, who is really in the hot seat on this -- and Linda McMahon in operations -- for us to tell what's really working and what's not, because we think we see patterns, you know, where one state is falling short of another, and the response, invariably, and understandably, is, well, you know, it's apples and oranges because Florida is measuring it differently from Virginia, or vice versa.

And what we've tried to do -- we had an effort to do this once before, and it

failed because it was too top-down federal. What we're trying to do now is really try to take the whole notion of partnership seriously and get all the states involved and talk out the issues in advance and really make sure we come up with a plan that they want to do.

And so we had about 250 people -- 250 people together for three days, and we had a couple of large kickoff sessions, and then we broke out into small groups, and then we reconfigured them, and we're still working on that.

But I think that you're right; if we're -- particularly when we're dealing with the bigger picture issues, we do have to pull out of our day-to-day and we have to sort of engage with sometimes a very different group of people, and think about things differently and try to make sure that for all our very careful attention to the detail, you know, we haven't lost track of the big picture.

So that's what we're doing

tomorrow. That's what we did last week with the state DDSs, and that's really what we're trying to do here -- not only today, but we've built in that this is not a one-shot deal; this is going to be an ongoing quarterly thing. It won't be rare diseases every time. You probably won't have another one for a while. We've got -- oncology is next. I think traumatic injuries after that, and then chronic disease after that. So there may be some overlap for some of you in the chronic disease area.

And then we'll have another four next year. So I think we're trying to take that to heart. You know, we may not take it to heart as much as we should in some areas, but we are trying to take that to heart.

But that was a fair question. I'll take a question like that anytime.

Anybody else? Going once. Going twice.

All right. Again, thank you all

very much. Thank you to my long-suffering staff who did such a great job putting this together, all the participants, National Organization For Rare Diseases, NIH, which has been invaluable, and anybody else that I've left out. This has been a terrific couple of days, and I thank everybody.

(Whereupon, at 3:26 p.m., the hearing was concluded.)

CERTIFICATE OF REPORTER

I, KATHY SAVICH, RPR, do hereby certify that the testimony that appears in the foregoing transcript is the testimony of said Witnesses, were taken by me in shorthand and thereafter reduced to computerized transcription under my direction; that said transcript is a true record of the testimony given by said witnesses to the best of our ability; that I am neither counsel for, related to, nor am employed by any of the parties to the action; and further, that I am not a relative or employee of any attorney or counsel employed by the parties thereto, nor financially or otherwise interested in the outcome of the action.

Kathy Savich, RPR

Court Reporter