

**Testimony Submitted by the National Organization for Rare Disorders
Before the
Social Security Administration
Public Hearing on Compassionate Allowances
December 4, 2007**

Good morning. My name is Diane Edquist Dorman, Vice President for Public Policy for the National Organization for Rare Disorders. On behalf of NORD and the millions of Americans affected by rare diseases, I want to thank you for the opportunity to speak before you today regarding the Social Security Administration's desire to improve the rules for Compassionate Allowances for individuals affected by a rare disease.

As mentioned in our comments submitted to the Social Security Administration this past September, the men, women and children seriously affected by rare diseases, many of which are severely debilitating and/or life-threatening, are routinely denied Social Security Disability insurance and are forced to go through the lengthy and often expensive appeals process. This is because nearly 100 percent of the rare diseases are not in the *SSA Listing of Impairments*, nor are they included in the U.N's *International Classification of Diseases*.

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

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

- According the Office of Rare Diseases at the National Institutes of Health, there are approximately 7,000 known rare diseases, each of which affects far fewer than 200,000 people in the U.S. The NIH estimates that, in the aggregate, between nine and ten percent of the U.S. population has been diagnosed with one of these rare diseases, disorders or syndromes.
- Eighty-five to 90 percent of known rare diseases are chronic, serious or life-threatening.
- Approximately 80 percent of these diseases are genetic. Consequently, I make the assumption that children are inordinately impacted. This is evidenced by the fact that about 50% of the over 300 orphan products are approved for pediatric use.

- The diagnosis of a rare disease often takes years. Little evidence is available to support a diagnosis. Experts in the field are few and far between, and doctors know little of these diseases. As a result, patients and their families are shuffled from one specialist to another.
- In a survey NORD conducted with Sarah Lawrence College several years ago, 42% of the respondents said they were prevented from working because of their disease. (Survey sent to 15,000 people. Response rate was 9%)
- In same survey, 77% of the respondents said their rare disease had caused them or their families a financial burden. 32% characterized that burden as “extreme.”
- Many rare disorders have no surrogate endpoints, markers or tests. Some are diagnosed based on clinical observations only, others by genetic tests.

Before closing, I would like to share with you, in their own words, some of the difficulties that have been described to me by leaders of NORD member organizations.

Guillain-Barré Syndrome

Guillain-Barré Syndrome and its variants (such as CIDP) are rare neurological disorders which strike many patients with paralysis for the short term. If these people could get on disability at the earliest time after diagnosis, it would help them to recover and, in many cases, go back to their previous lifestyles – including work. With GBS and CIDP, the vast majority of patients are unable to function at full-time work from the onset of the disease and for a period of at least one year, even with full medical compliance and physical therapy.

Lymphangiomyomatosis

LAM is a serious lung disease that tends to strike young people, almost always women, and is thought to be in some way related to hormones. It is not caused by smoking. A woman in her 40s told us that she received the diagnosis in December of 2005 and was told by her physician at Cleveland Clinic that she would no longer be able to work. She had previously worked as a clerk in retail stores. The woman first applied for assistance in January of 2006, but a few months later received a denial letter which stated that she should still be able to do "sedentary" work. She hired a lawyer and appealed the decision. Again, she was denied. She hired a different lawyer and appealed again. This time, she was notified (in November 2007) that the denial had been reversed. By that time, she had lost her home and her car. She was getting food from a food bank, and

relying on friends and people from her church for help with buying necessities, such as toothpaste. Worst of all, she had accumulated \$3,500 in attorney fees that wouldn't have been necessary if her physician had been better able to make his voice heard earlier in the process.

Late-Onset Tay Sachs

Case 1 - My main concern at this point with the SSA is that they are constantly asking us to prove that my sons are truly disabled. I have provided documentation regarding the fact that LOTS is a degenerative disease and the condition will not improve.

Case 2 - Because I have four disabled children, I realize that my experiences will be multiplied many times over. There have been many times during the past 15+ years that I have been reduced to tears because of the callous treatment that my sons and I have received.

Case 3 - I understand the system was designed to care for individuals like Laura. Why then is it such a paperwork nightmare – repeated yearly to get and keep SS and Medicaid when she has absolutely no hope of recovery and everyone in the medical profession knows it?

Marfan Syndrome

“On November 3, 2004 another member of our group lost her fight with Marfan syndrome and believe me she fought very hard. She was 39 years old. She has spent the months since February 2004 trying to prepare her son for her ultimate death, make arrangements for him to be taken care of after her death and try to get social security disability so she could take care of him while she lived. Just last week, social security denied her, for the second time, any benefits. Honest to Pete, I don't see how they could do that. If they read her medical records and learned she still had a dissection that was not repaired at the time of the emergency surgery, that she could not walk even 15 feet without becoming totally out of breath, that she bled into her right leg if she stood too long, she had suffered a heart attack, and she had Marfan Syndrome; how could they deny her. Please someone explain to me how the blazes they could turn her down. This woman had no means to provide for her son and herself except for the goodness of other people. She tried to work and just could not. Up until the time she had her emergency dissection she worked manual labor jobs such as hand unloading semi's at a distribution warehouse to take care of her and her son. She did this because no one ever told her that she should not lift heavy things, especially not repetitive. Now, I hope her family sends her obituary to the person who wrote the denial and asks them what might it have been that they missed in her request for help. It just does not seem that our system of helping people is working. Even though I am not a very outgoing person, I would like to look the people in the eye who made the decision and ask them how they are going to pay for her to be buried and how this little boy is going to make it with no mother and no father. Yes, he will be staying with an aunt but that is not his mother.

Reflex Sympathetic Syndrome

Reflex sympathetic dystrophy syndrome (RSDS), also known as complex regional pain syndrome, is a rare disorder of the sympathetic nervous system that is characterized by chronic, severe pain. Excessive or abnormal responses of portions of the sympathetic nervous system are thought to be responsible for the pain associated with reflex sympathetic dystrophy syndrome.

The Reflex Sympathetic Syndrome Association of America conducted an on-line survey in 2005 with the assistance of the Johns Hopkins School of Medicine. Included in the survey (n=1362), were three questions about Social Security.

Did you ever apply for Social Security Disability Benefits?

	# Responses	% of Group	% of Total
Yes	666	48.9	48.9
No	504	37	37
Still in Process	192	14.1	14.1

Were you denied the first time?

Yes	424	63.7	31.1
No	242	36.3	17.8
Total of Group	666	100	48.9

Were you denied a second time when you applied for SSDI benefits?

Yes	283	66.8	20.8
No	141	33.3	10.4
Total of Group	424	100	31.1

47% of respondents had contemplated suicide; 15% had acted on impulse; average pain score was 7 (on a scale of 1-10); 60% were unable to work; only 16% worked full-time

Tourette Syndrome

For those with severe, disabling TS motor tics as well as inappropriate, prominent vocal symptoms, the quality of their lives can only be described as non-existent. Some of the most disabling factors that make it veritably impossible to cope with TS symptoms include self-mutilating behaviors, isolation, and employment discrimination. We suggest that SSA determine disability based on a case-by-case basis. These would include, but not limited to, diagnosis, symptom severity and impairment, as well as quality of life

Wegener's Granulomatosis

I have had Wegener's Granulomatosis for almost 13 years now, and in 2005 I had [a] major flare of the Wegener's, which required major hospitalization. I was in the hospital for two months at that time, and then three other times that year.

Anyway, I had applied for Social Security Disability , was denied, appealed, was denied again, and then in November of 2005, I was told to file with the judge, and right now, I have an attorney, but am still waiting for a hearing date to be set. They keep telling me that I should be getting a hearing date anytime now, but it is really getting to be frustrating, and would like to see something happen soon.

These are just a few examples of the types of frustrations, delays, and unnecessary expenses often experienced by people with serious and even life-threatening rare diseases who apply for disability assistance.

When Commissioner Astrue spoke at the NORD Annual Conference in September, he emphasized that the impetus for change comes from SSA and not from external sources. In other words, SSA truly desires to provide better service to claimants who are currently being subjected to unnecessary, emotionally draining, and costly delays. NORD applauds this pro-active approach and wants to assure all involved that we and our medical advisors will support SSA in the effort to improve its service for patients with rare diseases in any way that we can.

Thank you.

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