



September 17, 2007

Commissioner of Social Security  
P.O. Box 17703  
Baltimore, MD 21235-7703

Via Email: [regulations@ssa.gov](mailto:regulations@ssa.gov)

Re: Federal Register, Vol. 72, No. 146  
20 CFR Parts 404, 405, and 416  
Docket No. SSA 2007-0053  
RIN 0960-AG54  
Proposed Rules: Compassionate Allowances

Dear Sirs or Madams:

We are extremely supportive of SSA's initiative to improve its rules for Compassionate Allowances for people who qualify for Social Security Disability Insurance (SSDI) benefits. People who are seriously impacted by a physical or mental impairment and cannot work, deserve quick and humane decisions by SSA so they can focus on their health.

The National Organization for Rare Disorders (NORD) represents an estimated 25 million Americans with rare "orphan" diseases. The National Institutes of Health (NIH) says there are approximately 7,000 of these conditions, each one affecting fewer than 200,000 Americans (as defined under the federal Orphan Drug Act of 1983). There is no master list of all rare disorders. However, the NIH Office of Rare Diseases maintains a list of over 6,000 of these conditions. Some are severely debilitating and/or life-threatening, but some are not. Unfortunately, the United Nation's International Classification of Diseases (ICD) does not list most of the rare disorders.

SSA's sequential evaluation process is logical for the most well-known and well-understood conditions. Unfortunately, because most rare disorders are not on the SSA Listing of Impairments, many people with rare disorders are ordinarily delayed or denied benefits and are forced to go through the appeals process.

In answer to your questions in the Federal Register notice:

- ✓ Creating a comprehensive list of rare disorders for use by SSA is an exciting proposal. However, simply naming the disorder (e.g., the NIH list from the Office of Rare Diseases) will not be enough. You will need to have an explanation of the disease for use by SSA staff so they will be able to understand the disorder, the way it affects patients, and suggests the degree of severity a person must have before they can be judged as disabled. If these descriptions were readily available to SSA employees, they would be able to quickly make decisions about compassionate allowances especially for many applicants with life-threatening and degenerative rare disorders.
- ✓ Many rare disorders have no markers or tests that could quickly provide medical evidence of disability. Some are diagnosed based on clinical observation, others by genetic tests, etc. Most people with rare disorders go undiagnosed for extensive periods of time simply because there are no laboratory or imaging tests that would diagnose them. This is a very serious problem for these populations and their physicians, and we recognize it will be a major problem to SSA. Nevertheless, for those who do have measurable evidence (e.g., the size of tumors, blood or breathing tests, etc.) a lot can be done to help SSA create its list. In fact, it may be preferable for SSA to determine disability based on symptoms rather than diagnoses. For example, paralysis of an arm or leg can make a person disabled no matter what the cause or diagnosis is.
- ✓ Another problem will be standardized codes. There are no codes for most rare diseases so many patients are coded (for billing purposes) according to symptoms, not their diagnoses. Others are put in general categories such as "birth defects" or "genetic diseases." We support creation of a new SSA list of impairments based on established diagnoses. The list should be put on SSA's Website so patients and doctors can have access to it.
- ✓ We suggest that SSA should not rely on generic letters from doctors to determine disability. Rather, SSA should create a form for physicians with specific questions so that doctors will provide evidence that you ask for. The form should go on the Internet with some of the following questions:
  - 1) What is the diagnosis?
  - 2) Explain the disorder in detail.
  - 3) What are the patient's symptoms?
  - 4) Explain exactly how the disorder interferes with the patient's daily functioning.

This type of questionnaire should provide information that case reviewers need when the diagnosis is unusual.

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- ✓ The ICD and DSM are incomplete lists. We believe that NORD and its member agencies can locate medical experts on even the rarest diseases who can be consulted for development of a comprehensive list. Additionally, the NIH, CDC, and National Library of Medicine can also be helpful to SSA in creating the list.
- ✓ Keeping the list up-to-date will be essential. It should be an electronic list that can be updated quickly. New "orphan drugs" are making many untreatable diseases treatable. However, many of these people will remain disabled permanently or for a long time after a treatment comes to market because they won't be cured, and treatment will be chronic.

We hope our comments will be helpful. Please feel free to call on NORD any time in the future. Our medical advisors remain ready to help.

Very truly yours,

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