

Witness Questions
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- 1) With regard to the claims of individuals with rare diseases that unquestionably qualify under SSA's Listing of Impairments, what is your experience or the experience of the people you advocate for in filing for Social Security benefits? Are claims allowed quickly, or do they take longer than anticipated?

The experience of individuals with Tuberous Sclerosis Complex (TSC), an autosomal dominant genetic disorder that affects 1 in 6,000 live births, or approximately 50,000 Americans, in filing for Social Security benefits is mixed. Some individuals are able to apply and receive approval for benefits as quickly as 3 days later, whereas others are denied and then required to appeal. This is particularly true for adults with TSC who are applying for the first time as adults when their condition becomes so serious that they can no longer be employed and require SSA benefits.

- 2) In those cases where claimants' conditions qualify for benefits and it takes longer than anticipated for claims to be allowed, what do you think are the underlying problems? For example, are there people (including SSA's adjudicators, members of the medical community and others) that are unfamiliar with the names of rare diseases or how these conditions impact claimants? Is obtaining medical evidence a problem? If there are common errors, what are they?

The experience of individuals with tuberous sclerosis complex and their families is that no one has ever heard of the disease, and it very often is confused with tuberculosis. Individuals who apply for benefits are advised to supply as much documentation about the disease as possible with their 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 individuals with tuberous sclerosis complex. Individuals with TSC 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 familiar with the disease. SSA adjudicators are not familiar with TSC, and the individuals applying must supply documentation of the disease manifestations, and extensive medical documentation regarding their disabilities that are caused by the disease.

- 3) SSA currently has processes in place designed to expedite the processing of claims. One process involves terminal illnesses (TERI) cases under the Disability Insurance (DI) program and Supplemental Security Income (SSI) program – and the other involves presumptive disability payments under the SSI program. How are these processes working for individuals with rare diseases?

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 is most often considered a chronic disorder, not a terminal disorder. TSC can be a terminal illness because of the

multisystem nature of the disease, affecting the brain, heart, eyes, kidneys, lungs, liver, skin and virtually any organ in 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 for TSC, individuals with the disease are treated for each of the symptoms of the disease – seizures, autism spectrum disorder, brain, kidney, heart, skin, lung and/or liver tumors, learning disabilities ranging from mild to severe, and mental health issues including depression, bipolar disorder, anxiety disorder, obsessive compulsive disorder and attention deficit hyperactivity disorder.

- 4) With regard to claims that qualify under SSA's current listings, what suggestions do you have for improving the current system for individuals with rare diseases? For example, would access to a list of rare disorders describing their impact and the objective medical evidence needed to establish the conditions be helpful to adjudicators? Can you suggest new ways that medical information pertaining to SSA's functional criteria could help improve the adjudicatory process?

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 that 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, including TSC, 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 TSC may be mildly affected as a child and not require SSA 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 over the last 10 to 15 years such that now individuals who are mildly affected by the disease are being identified and diagnosed with the disease, whereas in the past only the most severely affected and disabled individuals were diagnosed. The mildly affected individuals may have significant disability from one aspect of the disease even though they are unaffected by other aspects of TSC. 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 the genes and the development of treatments that will lead to clinical trials, the landscape for treatment will also be changing. 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 non-profit organizations have volunteer Professional Advisory Board composed of knowledgeable health care professionals who could serve as a resource to the SSA as needed to assist in obtaining information about a specific rare disease.

- 5) Please tell us about other suggestions you have about how we can improve our service to individuals with rare diseases.

The Tuberous Sclerosis Alliance appreciates the opportunity to participate in this hearing, and applauds the SSA on this initiative to improve 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 non-profit organizations to health care 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 process.