Testimony Prepared by Amy Kirk, Coordinator of Family Services, Batten Disease Support and Research Association

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?

Many of our families have had to individually list out the conditions of their child's disease. 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 a SSA decision, we tell our families to explain the symptoms of the disease as a means of a diagnosis. When a SSA adjudicator reads, "seizure disorder, blindness, fine motor skill impairment, gross motor skill impairment, mobility disorder, etc" a quick decision from SSA is more likely. When the term Batten Disease is listed, there is always an explanation that needs to come along with it. For the sake of both the families and the SSA, we have recommended the individual symptom listing as a way to describe the diagnosis.

This, in itself, poses two problems. One, it is not a true diagnosis. Yes, the child has a mobility disorder, but the mobility disorder is a condition of Batten Disease. While we are not lying on the application, we are not presenting the entire picture. The second problem is that providing medical information for each of these individual symptoms can be tedious to all involved. Providing the SSA with a diagnosis of Batten Disease would make the process much easier and faster. Instead of searching for ophthalmology records, physically therapy records, neurology records, special education records, etc. to prove the diagnosis and pertaining conditions, a diagnosis of Batten Disease, in any of the acceptable standards, would prove to be much simpler.

In general, claims are made in an appropriate time frame due to the fact that we have taught many parents and families over the years how to more quickly expedite their claims. If these accommodations were not made, the process would be much slower and more tedious for families, medical professionals, and the SSA.

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?

Most of the general public and general medical profession is unfamiliar with rare diseases. Even in terms of specialists, like neurologists, the rare diseases are hardly ever heard about and much less seen. With over 7,000 rare diseases in the United States (NORD, 2007), is it that unlikely that medical professionals may not be as up-to-speed on

the specifics of a disease like Batten Disease? Batten Disease affects 2-4 out of every 100,000 births. Currently we have approximately 400 children in the United States with a diagnosis of Batten Disease. 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 an SSA 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 SSA to also be somewhat knowledgeable about the rare disease, even if it just means having it listed in a book.

Providing medical records is not the 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 of the disease. Being that Batten Disease is an inherited genetic disorder, pin-pointing 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 SSA benefits.

While collecting the medical information may not be hard, it is time-consuming. For children with Batten Disease, medical information can come from many different professionals and 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 and complicated neurological diseases, the team of specialists can be large. Having to collect all this information is important though, to determine the appropriateness of the SSI eligibility and to give non-experts a better over-all picture of what the disease entails.

One of the most common errors, as all of you 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, respond with, "we are very over-loaded right now." While the SSA 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 large manual as a means of determination.

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?

This program isn't working because it isn't being advertised. This is the perfect type of program for people with rare diseases. However, it takes education and outreach on the part of the SSA to let rare disease organizations know about special clauses like these. Parents of children with rare diseases are too wrapped up in the present time with what is happening to their children right now, what symptoms do they display right now, that the thought of their child's life's end is not in the forefront of their mind. It may also be

interesting to know whether SSA workers and adjudicators are offering up these types of programs to parents with a child who has a rare disease. As stated earlier, we encourage our parents to write every symptom and condition of Batten Disease. While the symptoms are numerous, they do not point to a terminal illness. Parents or SSA 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 is lacking.

Up until now, the SSA has not done much to respond to and connect with the rare disease community. Even though rare diseases seem like a small percentage of the country's population, there are 25 million people that have a rare disease, not to mention the families that are also affected (NORD, 2007). With this new attention being given to the rare disease community, I encourage you to take it one step farther and begin providing the outreach and education about special SSA programs that would assist families and individuals experiencing a rare disease.

Both the TERI program and presumptive disability payments are very helpful. As a rare disease organization, we will continue to inform our families about these programs.

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?

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 "Batten Disease-Batten Disease Support and Research Association 1-800-448-4570," could help to answer any further questions an adjudicator may have without specifically talking 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 the 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 10 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. This is why it is so 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 wording as listed in the manual, yet it was the exact same disease.

Within the manual, it is also important to list all of the accepted means of diagnosis for the disease. Consulting with a rare disease organization or a rare disease's top researchers will provide you with all of the accepted means of diagnosis. In the case of many rare diseases, gene location for the disease has not yet been determined. There are other ways of diagnosing a disease without DNA confirmation. While those with Batten Disease strive for a DNA diagnosis, it can take a very long time to get. A child's SSA benefits should not be held up due to the inability of science to locate his/her defective gene.

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

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 the disease.

In the case of Batten Disease, as is 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 generic prescription drug lists that many pharmacies are allowing. Medications can range upwards of $300 for a month's supply—and that's only one medication!

It is also very hard for parents of children with a rare disease to both 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 families experience is the classic line, "you make too much money," and the SSA ends the communication. If the SSA cannot help a family, the workers need to be trained on what other avenues families can turn to for help. When it comes to a child with a rare disease, help is not easily found and having doors slammed in their faces does make the situation any easier.

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 SSA could also provide a rare-disease hot-line where families could call to ask questions or see if their rare disease is listed in the manual. They would also know the SSA's preferred term for the rare disease and the specific diagnosing criteria needed to qualify for benefits.

I want to take this time to thank the Social Security Administration for inviting the Batten Disease Support and Research Association to speak at today's hearings. I hope my comments and suggestions have proven to be helpful. I look forward to working with the Social Security Administration and the rest of the rare disease community in the future.

References

National Organization for Rare Disorders. (2007). http://www.raredisease.org.