SSI – the Key to self sufficiency and portal to additional services for the disabled.

Our son Keith has a rare, life-shortening, genetic disorder called Friedreich's ataxia. Though Keith was born a healthy infant in December 1985, by the time he was eight years old and in the third grade, he began to stumble and fall for no apparent reason. He also began to have increasing difficulty writing legibly. His hands were cramping and so were his legs. These symptoms worsened to the point that, when Keith was eleven years old, we had him tested and he was genetically confirmed to have Friedreich's ataxia. Like most parents who receive this diagnosis, we had never heard of Friedreich's ataxia. We learned a lot about the disease, though, on the day of diagnosis.

We learned that Friedreich's ataxia is a neuro-muscular disorder inherited by way of a recessive genetic mutation carried by about one in 90 people. We learned that, when two such "carriers" have children, each offspring has a one-in-four chance of inheriting the mutation from both parents and being afflicted with the disorder. We learned that Friedreich's ataxia is relentlessly progressive and would soon deprive Keith of strength and coordination in all four limbs, placing him in a wheelchair in his teens. We learned that Keith's vision, hearing and speech would all be diminished; that he would develop severe scoliosis (curvature of the spine) requiring surgical implantation of metal rods to straighten it; that he would be at greater risk of developing diabetes, and that he would most likely have a severe heart condition. Finally, we learned that there is no treatment or cure for Friedreich's ataxia and that average life expectancy for Friedreich's ataxia patients is early adulthood.

Within a few weeks of Keith's diagnosis, after a number of medical examinations, we knew he already had the severe heart condition and the early stages of scoliosis. His weakness and incoordination progressed rapidly and he began using a wheelchair full time when he was 16. His scoliosis also worsened and he required the spinal fusion surgery to correct his spine that same year. His vision, hearing and speech steadily declined and continue to do so. He developed type 1.5 diabetes at age 20.

So, by the time Keith turned 18 and was eligible to apply for SSI as an adult, he had been in a wheelchair for more than 2 years, had endured spinal-fusion surgery, could not speak clearly, and his vision and hearing were impaired. He had an Individual Education Plan for the final 7 years of his education. Keith had worked hard and done well in school, despite his disabilities, and graduated from high school with his class after posting a 4.0 GPA during his senior year. Although he went to our local community college for several semesters and did well in his classes, the relentless progression of his disorder left him extremely fatigued and made it increasingly difficult to read and keep up with his assignments. Transportation was another complicating factor in that Keith had never been physically able to drive so had to rely on others to get him around. With his physical abilities in a steep decline and the demands of his education on the rise, Keith reluctantly decided he could not continue his schooling. He was also disappointed to find that he was unable to find a job.

Again because Friedreich's ataxia is relentlessly progressive, Keith's physical capabilities have continued to decline steadily to the point of needing around-the-clock assistance for even the most basic needs. The diabetes associated with Friedreich's ataxia carries its own additional requirements in this regard. Keith is unable to perform the blood-glucose testing or administer his own injections, both of which are required several times each day.

With all of these increasing challenges, you can understand how welcome to our family was the very quick SSA decision on Keith's disability claim. You can imagine how encouraged Keith and we were when he received his first SSI check even before he received his approval letter! With no other income or resources, Keith had no other way to support himself. The SSI monthly payment helps Keith pay for rent, food, utilities and personal care items. It is critical for Keith to have the financial ability to take care of his basic needs.

Fortunately for Keith and us, Friedreich's ataxia is one of the rare disorders listed in Section 11.17 of the Disability Evaluation Under Social Security (Blue Book- June 2006). http://www.socialsecurity.gov/disability/professionals/bluebook/11.00-Neurological-Adult.htm. Because of Keith's experience, he and we now realize that Not only is the SSI program extremely important in and of itself, SSI approval also provides a gateway to applying for additional social services. For example:

Medicaid: Because Keith was approved for SSI, he was able to apply for Medicaid through the Commonwealth of Virginia. He met the federal income and asset eligibility standards and Medicaid helps pay for his doctor and hospital care, medication and durable medical equipment such as a wheelchair.

Home Based Care: Keith has continued to decline dramatically in his ability to take care of himself-- personal grooming, dressing, meal preparation and eating – just to name a few tasks he now finds very difficult or impossible. With SSI and Medicaid approval in place, Keith was evaluated and found eligible for Personal Care Services. He receives caregiver services for five hours per day, Monday through Friday. The caregiver helps him bathe and dress, prepares his meals, washes his clothes and vacuums his room. Not only is this of benefit to Keith, it allows us to a daily block of time for our employment or time to take care of the family's other routine needs.

Respite Care: When Keith's caregiver is not with him, either his mom or I have to be available to take care of his needs. This reality has become part of our daily lives. We love our son and want to help him in any way we can, but we realize, like so many others, we will not be able to help take care of Keith unless we also take care of ourselves and our other obligations. Keith and we were grateful, therefore, that Keith was granted approval of pre-authorized service for respite care in the amount of 720 hours per year. Respite care helps to refresh us, to sustain ourselves and our family. It helps us take care of ourselves and our family so we can better help take care of Keith.

I doubt that anyone will come before you and say that it is easy living with a rare disease or helping care for someone with a rare disease. I would like to assure you, though, that the SSI program helps many people, including our son, both directly and indirectly – in tangible and intangible ways. It helps directly by providing them the wherewithal to provide for themselves as much as possible. It helps indirectly by providing a valid foundation that opens the door to important additional social services. It helps in the tangible, physical ways I have outlined. It helps in important intangible ways, too. It reinforces the recipient's sense of self-sufficiency, self-esteem and dignity. It works in all these ways to help provide the recipient with a higher quality of life than could otherwise be achieved.

I applaud the Social Security Administration in reviewing the need for compassionate allowances in an attempt to streamline the system for other patients with rare diseases and I look forward to assisting in any way I can. In terms of your effort to develop the most effective methods for identifying rare medical conditions appropriate for compassionate use allowances, I suspect that you are already engaged in most, if not all, of the following undertakings, so I will simply emphasize them from the vantage point of our own, very positive experience:

1. I would suggest, for example, working closely with your colleagues at the National Institutes of Health (NIH). More specifically, I would suggest working with the NIH Office of Rare Diseases as well as with each of the NIH Institutes responsible for each of the rare diseases you review. I had the good fortune to serve for four years on the National Advisory Committee of one of those NIH Institutes – the National Institute for Neurological Disorders and Stroke – and would like to join millions of others in applauding the NIH as the world's leading funding agency for medical research. You are, no doubt, finding at the NIH highly qualified scientists who can help identify the "trigger points" within rare diseases that can serve to validate speedy SSI approval. If there is anything you believe a patient advocate like me might be able to do to be of assistance in this regard -- in a liaison capacity, for example -- I would be happy to work with SSA in any way that might prove helpful.

2. As President of the Friedreich's Ataxia Research Alliance (FARA), I would also like to suggest that you work with non-profit advocacy organizations that support research and services for patients with rare diseases. Such organizations have formed around many rare diseases and would be happy to help where they can. The National Organization of Rare Disorders (NORD) would also, I'm sure, be happy to offer assistance. To illustrate the point using our own organization, FARA, we frequently answer questions and provide advice to patients who are preparing to file an application for SSI. We help them, for example, in identifying the employment, education and medical documentation required for filing; with the specific Social Security regulations pertaining to Friedreich's ataxia, and in using the BEST online screening tool. Rare-disease patient advocacy groups tend to know a lot about their disorders, partly because they are accustomed to helping educate their physicians who, in many cases, learned only a small amount about rare diseases in their medical training. Such advocates and the scientists with whom they are closely aligned could offer significant help in identifying

the “trigger points” for compassionate allowances in these rare diseases. Again, the NIH and its Office of Rare Diseases are well connected with non-profit advocacy organizations in the rare disease community.

Thank you very much for the benefits the SSI program provides to our son and our family as well as to others suffering with Friedreich’s ataxia. Thank you, also, for your efforts to make SSI available quickly to the other families living with such devastating rare diseases through the proposed Compassionate Allowances Initiative.