Raising a child that is affected by a rare, life-threatening, chronic disease process is the training ground for one's endurance. You are required to be determined, tenacious and capable. During the early years affected families must wage a war on a nemesis that does not have a name and journey through the isolation of diagnosis purgatory until you are lucky enough to finally arrive at a diagnosis. According to a 2004 survey for our affected families it took an average of three-years to obtain an accurate diagnosis. During the period that it takes to arrive upon a diagnosis parents one quickly learns their child's very existence requires the parent to be stronger than they ever imagined possible. Self-pity is not a luxury one can afford to indulge. Therefore we are required to transform from the role of a helpless bystander into a capable and empowered advocate. This characteristic is fostered in our children who have a life-threatening rare disease.

The greatest fear of any parent is the loss of a child. The thought is banished before it can even before that agony can be imagined. When you do lose a child the luxury of denial no longer exists especially when you have another child who is affected by the same disease process that claimed his brother's life. When I lost my son in 1990 I was determined to do everything in my power to not endure that agony again. I set out to save his brother.

I was challenged to redefine normal. As we prepared for having a child we read the books on developmental milestones and planned a future for an able child. We didn't find the book entitled "The joys of raising the chronically ill child." However we did find the joys in the smallest of victories and we rejoiced in each milestone achieved. In the years leading up to adulthood we reared our son to be capable, not to be defined by his disease.

The lack of familiarity about Barth syndrome in the medical community is a topic for another testimony. In short the under-diagnosis of disorder affects outcomes. Co-morbid disease aspects are not appreciated and the biochemical basis of the disease is seldom if ever considered. The medical experts who have spent years studying Barth syndrome readily confess there is no way to determine the outcomes and long-term impact of the disease. There is no distinct genotype – phenotype severity relationship. It is not uncommon for children in the same family to experience varying degrees of systemic involvement. The symptoms can arise and claim a life in a matter of hours or the individual may experience debilitating pain and fatigue over the span of an all too brief lifetime.

The biochemical basis of the disease was first observed in individuals with Barth syndrome in 2000. Experts in lipid research are frequently baffled that someone could survive with such an abnormality. Protocols for labs to establish a diagnosis are not standard. The process to obtain an accurate diagnosis is subject to technical expertise in protocol and parameters of interpretation. Some symptoms that have been observed remain as anecdotal reports, not yet reported in medical literature.

There is one common truth… One does not recover from a disease causing genetic mutation. Individuals who have Barth syndrome frequently experience recurrent bouts of

diminished health. The question is not if the disease will manifest recurrent symptoms but moreover a question of when. Until the disease causing mutation has been treated the individual is not healed. Recovery is temporary. Therefore recovered is not the most appropriate term to describe times of well-being. Moreover this is nothing more than a recession until the symptoms manifest again.

The degree of risk cannot be predicted nor should the risks be discounted. Individuals with Barth syndrome are new survivors. Those of us who have older children who have survived into the second decade of life are the pioneers of uncharted territory. We are capable and have strived to raise able children to be independent assets to society. Childrearing for a chronically ill child affected by an orphan disease is challenging.

Not since 1988 when my children first fell ill to a nameless disease have I felt so powerless than I did when I received a denial from the Social Security Administration. How could someone who has never seen my son; or for that matter never seen anyone with Barth syndrome have the right to deny my child access to Social Security benefits? The term orphan disease never seemed so appropriate than the day the letter of determination arrived. Self-reported questions are difficult to answer and laden with bias.

These questions negate my son's ability to adapt. My son thinks he walks rapidly because we walk with him. He has never participated in competitive sports. Over the years we have designed activities that did not highlight his shortcomings. He did not graduate from high school until he was 20 years of age because his fatigue would not permit him to attend school full time but he did finish high-school. He walks up steps but each step is taken slowly, bracing his legs as he walks and pulling up on the rail.

Decisions should not be left to ill-informed reviewers. When expertise is required to make a diagnosis, treat the disease and carefully monitor progression how can an adjudicator be qualified to determine eligibility of a claim of disability without seeking expert advice? Disease processes vary in severity on a daily basis. One could interpret the explanation of denial for benefits for my son as 'You're sick but you are not sick enough'. Employers are not inclined to hire someone who will be there for work when they can.

Please provide a listing of rare diseases and the impact of the disease processes. Allow organizations such as ours to assist SSA to improve these processes. Please don't dismiss the health advocacy group's efforts to this underserved population. Please don't penalize someone for adapting to their life-long journey with a chronic illness. Their determination to be capable does not make them able. Their disability should not diminish their hope for a future or limit their opportunity to pioneer a new kind of normal in their life.

Testimony from Shelley Bowen, Barth Syndrome