

## Public Comments

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Before the

Advisory Committee on Heritable Disorders in Newborns and Children

October 1, 2008

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Dr. Howell, Ladies and Gentlemen of the Committee,

Thank you for the opportunity to address you today. My name is Ron Bartek. My 22-year-old stepson Keith has a neurodegenerative disorder -- Friedreich's Ataxia. Keith was a beautiful, apparently healthy child at birth and until about age 9, at which time he was doing well in school and was typically active. He was enjoying karate lessons and riding his bike with his brothers and friends. Keith's world began to change rapidly and drastically at that point, though, and by age 11, he was diagnosed with Friedreich's ataxia as his incoordination, cardiomyopathy and scoliosis became apparent. By age 16 he was unable to walk and his scoliosis required surgical implantation of metal rods along the length of his spine. Now, at 22, Keith has developed diabetes, is full time in his wheelchair, and is dependent on others for most activities of daily living. Although his intellectual capabilities remain intact, he has significant communication difficulties due to vision and hearing loss as well as slurred speech. He had to terminate his college education as a freshman due to difficulty overcoming all these challenges. We estimate that there are 5000-6000 individuals like Keith living in the United States.

On the day in 1997 that my wife Raychel and I received Keith's diagnosis, we learned three things: first -- Friedreich's ataxia has a horrific prognosis; second -- there was no organization focused entirely on supporting research and education in Friedreich's ataxia, although there were other organizations such as the Muscular Dystrophy Association for which Friedreich's ataxia was one of many disorders in their portfolio, but third -- the disease gene and molecular defect had just been identified in the previous year. With the support and encouragement of Drs. Giovanna Spinella and Audrey Penn of the NIH's Neurological Institute, we decided to form an organization to support research and education in this disease. I am co-founder and President of that organization -- the Friedreich's Ataxia Research Alliance (FARA) -- and on behalf of FARA and the patient and research community it represents, I would like to express our gratitude for the important service you provide in helping the most vulnerable members of our society.

Friedreich's ataxia is the most common form of inherited ataxia. About one in 50,000 people in the United States are born with this disease. Symptoms of Friedreich's ataxia include: progressive loss of strength and coordination (ataxia) in all four extremities leading to loss of ambulation within 6-10 years of onset, life-shortening cardiomyopathy, severe scoliosis often requiring surgical intervention, and type 1 diabetes. Onset of symptoms can vary from childhood

to adulthood. Childhood onset of Friedreich's ataxia is usually between the ages of 5 and 15 and tends to be associated with a more rapid progression and premature death.

Since the identification of the disease-associated gene and mutation in 1996, our scientific community has made tremendous progress in understanding the pathophysiology of the disease and function of the protein, frataxin, which is dramatically reduced in our patients. Frataxin functions in the mitochondria and is critical for iron metabolism, antioxidant protection, and overall energy production. A number of promising therapeutics are in clinical trials or in development. At present, in the United States and Europe, there are two ongoing Phase III clinical trials of the antioxidant Idebenone. A Phase II study of the iron chelator Deferiprone is also underway in Europe. These trials will be completed in 2009. Data from previous studies of both drugs suggest improvements in cardiac and neurological function that would, at a minimum, slow the progression of the disease significantly. This past July, Canada granted conditional approval for use of Idebenone in Friedreich's ataxia and FDA filing for approval is anticipated late in 2009. In addition, there is a Phase I clinical trial now underway in the United States of another novel antioxidant in Friedreich's ataxia. This and a number of other drugs now in development with our pharmaceutical partners hold promise for substantially greater benefit.

Due to the progressive nature of this disease and the strong likelihood that damage begins long before symptom onset, we believe that the earliest intervention with these treatments will be critical in reducing and preventing morbidity and mortality. Our organization is clearly not alone in this conviction. For example, at our 3<sup>rd</sup> International Scientific Conference on Friedreich's Ataxia, in November 2006, Dr. Story Landis, Director of the NIH's Neurological Institute, told FARA that her Institute was so encouraged by our progress and confident that we would achieve effective treatment soon, that we should begin working with NIH colleagues and others on the development of a newborn screening test. Since then, we have been working with a team of investigators at the Mayo Clinic in Minnesota led by Drs. Devin Oglesbee and Grazia Isaya to develop, using dried blood spots, a newborn screening test that is protein-based and multiplexable. The goal is to validate this test analytically and clinically with a pilot study beginning in 2009 of approximately 70,000 newborns so as to meet the nomination standards set by this committee. We would welcome in this development process the involvement of other academic groups and industry as appropriate.

We understand the role of this committee in ensuring that suitable screening tests are developed and safe, effective treatments are available for implementation in the newborn period. We also recognize the social, emotional, and ethical challenges of diagnosing pre-symptomatic individuals.

We look forward to the opportunity to keep you informed as we achieve critical milestones in test development and treatment outcomes. On behalf of Friedreich's ataxia patient families and our research community, thank you for your commitment to the health of newborns and children, for your attention and for allowing me to make this presentation to you today.