Ilyce Randell's testimony before the NIL, HHS, NICHHD

February 11, 2004

Thank you all very much for taking the time to meet with me today. I am the President and Founder of Canavan Research Illinois, a public charity with a mission of curing a fatal childhood illness called Canavan disease. My six-year-old son Max suffers from Canavan disease. Max has come to represent the hundreds of other children afflicted with CD as the "poster child"due to the extensive experimental treatments he has received, and the amazing progress he has made as a result. Max is also the motivation behind my aggressive efforts to see funding for rare disease research increased. I work alongside many other families in this country and worldwide, creating awareness and educating people about Canavan disease, while continuing to raise private funds for research that can offer hope to our children.

My son, and many other children, have made dramatic quality of life improvements due to the cutting-edge research supported by our organization, as well as a pilot grant awarded to Dr. Paola Leone for 2.9 million dollars from NINDS - this grant will expire next year and private funding alone is drastically insufficient to maintain the laboratory and clinical trial at Cooper Hospital in Camden, NJ. That is why I am here today. As in the past I am again asking for help and guidance from legislators in order to secure continued funding for Canavan disease research, without which our progress will be halted and all future gains lost.

I have two goals to accomplish during my visit to Washington. First, I am here as an educator to increase awareness about Canavan disease and its impact on society. As a person affected by an extremely rare disorder, it is my duty and obligation to help other people realize and understand that a disease they have never heard of, may one day affect them. It is a commonly held belief that research into rare diseases can offer insight and knowledge valuable to the study and understanding of more common illness that affect millions of people around the world. The pain felt and hardships endured by families affected by Canavan and other rare diseases are ours to bear alone, but knowledge gained from the study of these diseases could one day affect every living person as mysteries of the brain are discovered by our researchers.

My second goal is to suggest that additional safeguards are necessary to ensure that every American has an equal access to medical treatment-regardless of how rare their disease is, or whether or not it's study will offer financial gains to others.

"Rare disease" is too broad a term because it includes diseases that affect up to 200,000 people. Researchers studying "ultra-rare diseases", which affect less than 500 people, are

at an unfair disadvantage when they submit grant applications because they are competing for the same funds as researchers studying diseases affecting close to a-quarter-million people. Diseases such as Parkinson's and Type I Diabetes are grouped with Canavan and Maple Syrup Urine Disease. I would like to work with policymakers to create separate funding for researchers studying "ultra-rare" Mendelian (caused by a single gene mutation) disorders affecting less than 500 people. Diseases caused by a single mutated gene can offer insight into other genetic diseases where several genes are involved. Researchers working on diseases that serve as models to other common disorders must have equal access to federal funds. Often times grants are submitted that have "fund-able scores" but due to budgetary constraints they do not receive the award.

Here is an example: Grant application FD-R-002372 submitted to FDA Office of Orphan Products Development in support of the clinical reagent for the Gene Therapy of Canavan Disease project. This application received a good score, 186. But it was not funded, even after three thorough reviews and only because of budgetary cutbacks. This was the only grant application submitted for the development of gene therapy reagent in humans, setting the standard for all future clinical trials on gene therapy.
Increasing Government spending to ensure that "ultra-rare" diseases are fully researched would serve the public health interest because the study of diseases caused by a single mutated gene will give all medical researchers invaluable insight into other more common illness. Canavan is a degenerative brain disease; MS, Parkinson's, ALS, and Alzheimer's are just a few examples of related diseases that cause immeasurable suffering to patients, and their families, and cost our country billions of dollars every year. Researchers could offer insight into these common diseases if they had sufficient funding to study the basic science and pathways of diseases like Canavan, where the cause (a single defective gene) is already known.

Thank you for your time and consideration, I appreciate it very much!

Sincerely,

Ilyce Randell

President and Director, Canavan Research Illinois