

SPINAL MUSCULAR ATROPHY FOUNDATION

(Testimony of Ms. Loren Eng before the House Labor, Health, Human Services and Education Appropriations Subcommittee on Tuesday, April 27th, 2004.)

Thank you for allowing me the opportunity to speak with you today. My name is Loren Eng and I am the president of the Spinal Muscular Atrophy Foundation. We are a non profit organization working to develop treatments for SMA and have committed \$20 million toward that goal.

I am also here on behalf of the SMA Coalition, which is comprised of non profit groups from across the nation. Led mostly by courageous parents, these groups have transformed SMA from being a poorly understood disease, to one that is miraculously on the verge of treatment. These organizations have provided hope and comfort to children suffering from SMA

Most of all, I am here as a mother, asking for your help in saving the life of my 4 year old daughter Arya who was diagnosed with SMA 2 ½ years ago. Like most people, we had never heard of SMA. We now know that SMA is the leading and #1 genetic killer of infants and toddlers. As the most prevalent motor neuron disease, it causes muscles to wither away...it is incurable, untreatable, cruel, and deadly.

Over 20,000 Americans are living with SMA; up to 1,000 babies are born with SMA each year; and over 7 million Americans are carriers of SMA. Often described as a genetic form of polio, or a child's version of Lou Gehrig's disease, it is as common as ALS, Cystic Fibrosis, and Muscular Dystrophy. Yet, until recent discoveries, it was poorly understood and misdiagnosed. SMA destroys the motor neurons that control muscles, thereby causing them to wither away. In severe cases, which are most common, the muscle weakness leads to tracheotomies, feeding tubes, respirators and usually death before the child is two years old.

My daughter has a milder case of SMA. She is now four, and but for one gene, my husband and I feel like we hit the jackpot. She is sweet, witty and irresistibly adorable. Each day she grows smarter and funnier, and more aware of the world around her. Over the past 2 ½ years, we have watched the brutal effects of this disease on her frail body. She has lost the ability to walk, stand, sit up, or even crawl. Despite this, she is the most joyful child I have ever known. Like thousands of parents in this country, my heart breaks daily as I helplessly watch this disease ravage her.

Through the last few years, my husband and I have benefited from meeting other SMA parents, their courage has provided us with strength and inspiration. This disease doesn't 'just' kill children, it destroys entire families. For most, the emotional torment and

overwhelming cost of care for their children ruins them psychologically and financially. Care of a single SMA child can cost tens and even hundreds of thousands of dollars annually. In the last year of life, the cost tops \$1 million - which is why it is estimated that SMA costs America billions each year. Despite their daily pain and hardship, parents have managed to organize and advance SMA research for many years during a time when the government was providing virtually no funding for SMA research. It is the work of parents that has given us the realistic expectation of a treatment for SMA within four years.

SMA has the potential to be one of the most remarkable medical success stories in recent history. 15 years ago, the science of SMA was poorly understood. The gene was discovered 9 years ago and a series of findings have catapulted SMA to the verge of treatment. Amazingly, a number of compounds have already been identified which appear to treat SMA and the first clinical trials are underway. More importantly, some of these promising treatments are already FDA approved and could be used in the near term. Leading scientists (including Nobel Laureate James Watson) have made an eloquent case to NIH Director Dr. Zerhouni that SMA has gone from being a medical mystery, to being a disease with a real chance of treatment in the near term. We are pleased that, NINDS, under the leadership Story Landis, is actively pursuing the scientific opportunity to treat SMA.

Given the tremendous possibilities that lie before us, it is critical that government invest funding to match the work of charities. The anguished parents of dying children are pushing as hard as they can to end this disease. I am here asking you to help support our work by ensuring that NIH receives sufficient funding and support to complete the development of a treatment.

In reality, this is about much more than just one disease and 20,000 children. SMA has become a shining example for the value of investing wisely in medical research. As a result of major scientific findings, we have turned despair into hope in just a short few years. However, SMA is also an example of what needs to be fixed in the system. NIH and government funding are simply not set up to move quickly and strategically. In today's world discoveries can come at a lightning pace. We need to ensure that the government can respond quickly by strategically identifying opportunities and acting upon them. Otherwise, patients' lives will continue to hang in the balance. We will also waste money – both the billions that have been invested in genetics research as well as the billions being spent on caring for patients. Far less money, invested shrewdly in developing treatments, will save both lives and tax dollars.

My daughter just started school last fall. While it has been exciting, she now sees kids running, jumping and playing. Arya realizes that there is something seriously wrong and different about her. My husband and I struggle to answer her questions which come fast and furiously. She has begged to know—“Why can't I do the things other kids can?” “Will I ever be able to run and jump?” “What is SMA and why did God give me SMA?” Last week, before she went to bed, she sobbed uncontrollably in my arms and cried in frustration, “I do not want to have SMA anymore!”

We can't bear to think of the day that we will have to tell her what the future holds for her. With your help we can tell her, and thousands of other families, that a real treatment for her disease is coming soon. Thank you.