Testimony of Susie Trotochaud of Georgia
Before the Labor, Health and Human Services, Education and Related Agencies Appropriations Subcommittee of the
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Parent of 12 year old twins with Usher Syndrome, Parent Advocate representing the Coalition for Usher Syndrome Research and individuals with Usher Syndrome

Good morning Chairman Kingston and Members of the Committee. Thank you for the honor of appearing before you today. My name is Susie Trotochaud from the state of Georgia. I am here on behalf of the Coalition for Usher Syndrome Research to respectfully request this committee encourage NIH funding of $20 million in FY2014 to promote more research into Usher Syndrome.

Usher Syndrome is the number one cause of deaf-blindness. Deaf-Blindness. Imagine sitting here unable to hear my words and unable to see me. Silence and darkness. In the United States, it is estimated that about 45,000 people have this rare genetic disorder. Two of them are my children, Cory and Joanie Dorfman.

Cory and Joanie were born 8 weeks early. Although they spent several weeks in ICU fighting to learn basic survival skills, like breathing and eating, these would not be their greatest challenges. Before they were released from the hospital, they were given a newborn hearing screening. It was determined that they were both profoundly deaf. As we struggled to understand what this meant and how this could have happened, I realized that they would never be able to hear me say “I love you” and I would never hear those sweet words from their lips. The sounds of our life, children laughing, singing, school plays, graduations, celebrations, were suddenly silenced.

Our heartache changed to hope when we found out about the cochlear implant. By 12 months, Cory and Joanie were implanted and began hearing their first sounds. By 1-1/2 years, they had said their first words, and by 3 years, we realized that they could be mainstreamed, go on through high school and even college, just like their peers. Although they would always have to work a little harder, the sounds of opportunity returned to our lives. And I remember my husband saying to me at that time, "At least they're not blind."

But about a year ago, that all changed. After my daughter entered a darkened hallway in a restaurant and asked me where the bathroom was, when the door was literally four feet in front of her, we became concerned. When she gingerly stepped down a pathway at night, seemingly feeling her way with her feet, we knew we had a problem. Many months of extensive testing and waiting confirmed what we, by then, already knew. Joanie had Type I Usher Syndrome. Reading the description of Type I Usher was like reading her biography: Born profoundly deaf, delayed development especially walking, balance issues, and loss of night vision beginning at around 10 years of age. What would follow would be loss of peripheral vision leading to tunnel vision, and eventually blindness. With no intervention, my 12 year old daughter will be blind by 20. And although my son currently has less visions issues, testing confirms he also has Usher. He may retain some of his vision into his 30s.

That's the thing with Usher. It strikes in varying time frames. Type I, like with my children, is characterized by profound deafness at birth followed by blindness in early adolescence; Type II individuals may have moderate to severe hearing loss followed by blindness; and Type III experience loss of hearing and sight throughout their lives. How quickly and how completely each person losses their vision also varies, but the way it happens is consistent. Night blindness, then peripheral vision is lost as darkness closes in on their sight.

Usher is a rollercoaster ride of loss, grief, adjustment, and loss again that never ends as one more setback always lies around the corner.

People with Usher Syndrome, like Cory and Joanie, have worked hard to overcome some of their hearing challenges by using cochlear implants, hearing aids, sign language and more. But how do you overcome the loss of sight? Think of yourself, sitting here communicating by sign, knowing that you are losing your vision, knowing you are about to lose your way of communicating with the world around you. Frightening, isn't it?

Like you, my hopes and dreams for my children have always been that they grow up happy, do well in school, attend good colleges, get meaningful jobs and give back to their community. But the reality we are facing is that 8 out of 10 deaf-blind people are unemployed, not to mention the physical and emotional hardships, the stereotypes of being deaf-blind, the loss of productivity and ability to do a job, ultimate depression, and perhaps even suicide.

Add to that the reality that our country spends an estimated $27 billion annually in care and support services for people with major visual disorders. That doesn't even include the costs associated with hearing impairment.

Those are statistics; people with Usher aren't. Since joining the Coalition for Usher Syndrome Research, I have spoken with or met dozens of people who are determined, focused, and working everyday to help themselves, their loved one, or in some cases complete strangers, figure out how to treat this syndrome. Usher genes are complex, long protein cells which require significant investment in research if we are ever to find a cure or treatment. We can't do it alone.

Through the Coalition, we have brought the Usher community and researchers together by:

- Establishing a registry of individuals with Usher Syndrome which is available for research or clinical trials at no cost. Our registry currently has families from each of the 50 states and 23 countries.
- Sponsoring annual family conferences, webinars and monthly conferences that provide information and support to all of those living with Usher.
- Paving the way for an International Symposium on Usher Syndrome Research in 2014 to develop a roadmap for future research projects to bring us closer to viable clinical trials.

With this in place, we have begun bringing brilliant researchers together who are working on developing treatments every day. Researchers like those in Oregon and Pennsylvania who are working on gene therapy treatments, one of which began clinical trials this year. Researchers in Louisiana, who have been able to rescue the hearing in mice with Usher Syndrome using a drug therapy that holds promise for rescuing vision, as well. Researchers in Iowa, California, Nebraska, Massachusetts, Florida, Texas, and many other states, who are collaborating with each other and with families through the Coalition to advance all kinds of Usher syndrome research.

But still this is not enough. My daughter, Joanie, will be blind within 10 years; my son, Cory, in 20. Jessica, a 17-year old with Usher, remains hopeful that something will help her retain her vision before she loses it at 30. Megan, a promising architect, has already altered her career goals as her vision has begun to slowly fade and every day she prays for something to help. Moira has lived well into her adult life working harder than everyone else to compete in a hearing and seeing world, but complete blindness is now taking away her ability to lip read and communicate with her friends and family.

We cannot help any of these people or the tens of thousands who have Usher or countless others that will be born in the future with this devastating genetic disorder without Federal support. There are dozens of different mutations that cause Usher Syndrome and the pace of research is slowed dramatically by the lack of researchers and funding. The infrastructure is there to find treatments, but the significant financial support is not. We believe that $20 million in support this year and an increase of that amount over the next several years would lead to viable treatments for those with Usher Syndrome within a decade. We are asking you to supply this last critical resource to help us find a cure.

When you review the report on categorical spending by the NIH, Usher Syndrome is not even listed. Rare diseases with similar incident rates average around $50 million annually. These investments have resulted in significant discoveries for these diseases, and there is reason to believe that we can see these same results or better for Usher Syndrome. The researchers are there, waiting to discover what we only dare dream of: An opportunity to allow deaf children and adults who are going blind, a chance to see.

I will leave you with the words of Helen Keller. "It is a terrible thing to see, but have no vision." I hope that this committee will have the vision to see the opportunities before them. Together, we can find a way to end deaf-blindness. I thank you on behalf of all those with Usher Syndrome, their families, and most importantly to me, my children, Cory and Joanie. I am happy to answer any questions you might have.