

AEVR/FFB Congressional Briefing Highlights Urgency for Deaf-Blind Research

Imagine trying to live with the challenges of combined deafness and blindness and, at the same time, being unseen and unheard by society. That's the unfortunate irony for the 45,000 Americans with Usher syndrome, a devastating inherited disease that robs people of their vision and hearing.

On February 5, AEVR and the Foundation Fighting Blindness (FFB) hosted a Capitol Hill briefing to make legislators aware of Usher syndrome and to support its increased research funding at NIH. The three NIH institutes currently funding research are the NEI, the National Institute on Deafness and Other Communications Disorders (NIDCD), and the National Institute of Child Health and Human Development (NICHD). Much of the research underway not only impacts people with Usher syndrome, it is provid-

Syndrome is a vicious disease." His goal, simply stated, is to add at least 20 more years of vision for individuals who are often blind by age 20, just when they are starting to live independent lives.

Usher syndrome is caused by a genetic variation (a mutation) in the cells of both the retina and the inner ear. Genes lead to the production of proteins, and if a defective gene leads to a single missing or incorrect protein, it can mean serious trouble. Dr. Kimberling explained that Usher proteins in the retina work together like a crane to deliver important chemicals from one end of a cell to the other. If one protein isn't doing its job, it's like losing the motor or the hook or the cable, and ultimately, then the crane doesn't work, and the health of the retina is severely affected. Dr. Kimberling added that much of what is known about

mates that as many as 15 percent of these hearing impaired children will eventually lose their vision. However, if children with Usher syndrome can be identified early on, through genetic and vision testing, then measures can be taken right away to preserve their vision as much as possible. The progress of vision loss could potentially be slowed by the administration of neuro-protective treatments, which are in clinical studies right now. Also, nutritional supplementation with fish oil and vitamin A appear to slow vision loss in some individuals. Even wearing sunglasses in bright outdoor light might give a person two or three years of additional useful vision.

Not only does vision preservation provide these individuals with a better quality of life, it is likely to give them the opportunity down the road to benefit from an emerging curative treatment such as gene therapy, which Dr. Kimberling calls "the gold standard" for treating Usher syndrome. Additionally, he noted, by keeping individuals independent and productive, we can save much of the \$2 billion that he estimates that Usher syndrome costs our economy annually in health care and other supportive services.

Dr. Kimberling emphasized that Usher syndrome research is greatly underfunded and the condition is greatly overlooked. "Few people have heard of the disease, yet it is more common than amyotrophic lateral sclerosis (ALS, or Lou Gehrig's disease) or Huntington's disease."

"My goal is to add at least 20 more years of vision for individuals who are often blind by age 20" Dr. William Kimberling

ing important information about how all people see and hear.

Featured speaker William Kimberling, Ph.D., one of the world's leading Usher syndrome authorities who conducts research at Boys Town National Research Hospital in Omaha, Nebraska, and at the University of Iowa in Iowa City, explained that the combined impact of hearing and vision loss is many times greater than deafness or blindness alone. "Deafness and blindness together is not like 1+1 = 2. It's like 1+1 = 10," he said, adding that, "Usher

Usher syndrome is relatively new information. "Up until about the year 2000, we had no idea that such a mechanism [the crane-like structure] existed." So far, variations in nine genes have been linked to Usher syndrome, and researchers know there are more to be identified.

One immediate goal of Dr. Kimberling's research is to provide inexpensive and accessible genetic screening for young children who are born deaf or with severe hearing loss to determine which of them is affected by Usher syndrome. He esti-



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Left to right: Moirra Shea, a member of the FFB board who offered a firsthand account of living with Usher syndrome, with Stephen Rose, Ph.D., FFB's Chief Research Officer. Said Ms. Shea: "The vision loss process is like a series of cliffs. You go through life, and there's a big drop in your vision, you adjust to it, move forward, and then there's another cliff," she said, adding that "I am in a constant state of grieving."



Left to right: William Kimberling, Ph.D. with Baldwin Wong from the National Institute on Deafness and Other Communications Disorders (NIDCD) and Andree Reuss from the NIH Director's office, detailed to NIDCD



Dr. Kimberling with Erik Fatemi, who works for Senator Tom Harkin (D-IA), Chair of the Senate Labor, Health and Human Services, and Education (LHHS) Appropriations Subcommittee



Left to right: FFB Chief Executive Officer William Schmidt with Mike Ryan from the office of Cong. Steve Israel (D-NY). Mr. Ryan's sister was recently diagnosed with Usher syndrome.



Dr. Kimberling with Cong. Dave Loebsack (D-IA), a long-time medical research supporter



Senator Ben Nelson (D-NE) offers Dr. Kimberling a bandage to cushion a blister he developed during his busy schedule of eight Capitol Hill visits. Senator Nelson and Senator Susan Collins (R-ME) led Senate economic stimulus bill negotiations that retained the \$10 billion funding level for the NIH.