

USHER SYNDROME
COALITION



9th ANNUAL USH CONNECTIONS CONFERENCE

July 15, 2017

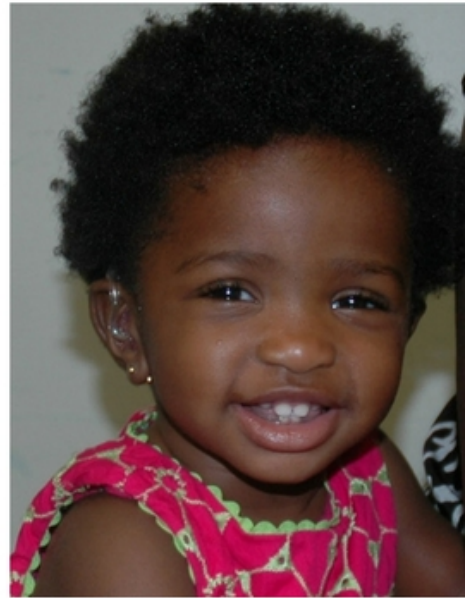
PROGRAM

Chicago Marriott Downtown Magnificent Mile | Chicago, Illinois



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***Walk in My Shoes* Book is Available!**

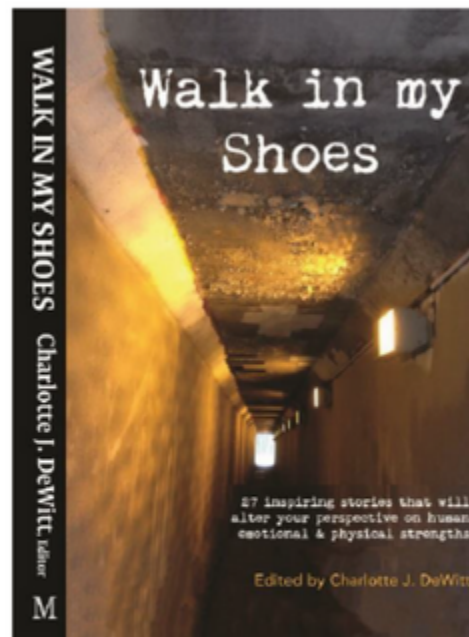
27 inspiring stories that will alter your perspective on human emotional and physical strengths

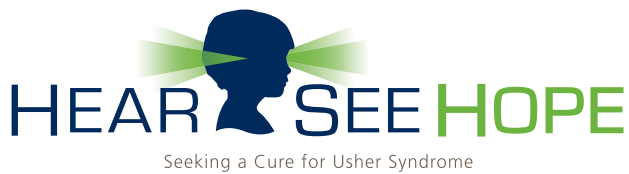
Walk in My Shoes is a book unlike any other. It is a unique collection of 27 powerful stories by individuals who have witnessed or experienced the tragedy of losing not one, but two senses: hearing and sight. The loss is caused by a rare disease called Usher syndrome. These stories will give you a glimpse into the world of the deafblind and their families and friends.

Where to order: Amazon, Createspace, Barnes & Noble, BookShare
Also available in Braille and Audio at Library for the Blind Center
Paperback ISBN: 978-1-945756-11-5
Ebook ISBN: 978-1-945756-12-2

***Walk in My Shoes* produced by Ramona Rice: rricetx@gmail.com**

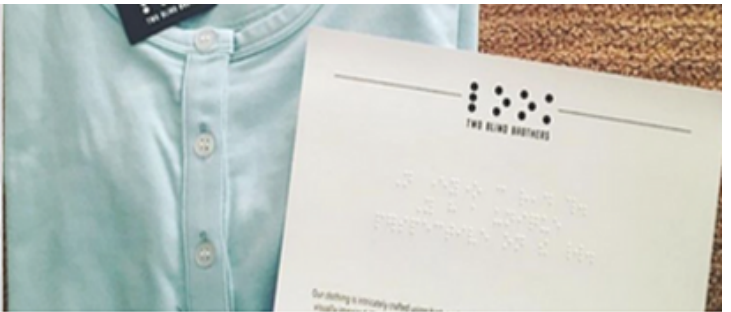
All proceeds from book sales are donated to the Usher Syndrome Coalition to help fund scholarships to the annual USH Connections Conference.





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USH Connections
Conference

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Saturday, July 15, 2017

SCHEDULE AT A GLANCE

7:00 – 8:30AM	Continental Breakfast and Registration Halstead and Marriott Ballroom, Fourth Floor
8:30 – 8:45AM	Welcome Address Mark Dunning, Chairman, Usher Syndrome Coalition
8:45 – 9:30AM	Keynote Speaker – Curing Usher Syndrome Edwin Stone, MD, PhD, Wynn Institute for Vision Research, University of Iowa
9:30 – 10:00AM	Life Without Limits – No Barriers Bill Barkeley, Featured Usher Syndrome Speaker
10:00 – 10:30AM	Break/Networking
10:30 – 11:00AM	Gene and Stem Cell Therapy for Usher Syndrome Ian C. Han, MD, Wynn Institute for Vision Research, University of Iowa
11:00 – 11:30AM	The Use of Antisense Technology for the Treatment of Usher Syndrome Michelle L. Hastings, PhD, Chicago Medical School at Rosalind Franklin University of Medicine and Science
11:30 – 1:00PM	Lunch/Networking
1:00 – 1:30PM	The RUSH2A Study: Gaining Insights into USH2A Ben Shaberman, Foundation Fighting Blindness
1:30 – 2:45PM	USH Research Panel Q&A
2:45 – 3:15PM	Break/Networking
3:15 – 4:45PM	USH Panel Discussion Moderator: Moira M. Shea Speakers: Lynne Murphy Breen, Linnea Haga, Derrick Phillips, Kevin Richmond, Diana Velarde
4:45 – 5:00PM	Closing Usher Community Photo Randall DeWitt, 'Usher Country' Rebecca Alexander, #Usher4UsherSyndrome Announcement
6:00 – 8:00PM	Evening Social Join us for food and fun at Lucky Strike! 332 E. Illinois Street, Chicago

WELCOME

Welcome to the 9th Annual USH Connections Conference, an event that has been described as "life-changing". For many, this will be the first time they meet others impacted by Usher syndrome. It will be the first time they hear from researchers working tirelessly towards a cure. For the first time, they will know that they are not alone.

As executive director of the Usher Syndrome Coalition, I have the privilege of connecting people. I get to bring people together and help bridge the gap between families and researchers. Thanks to your support, this vibrant community grows larger and more active every day. We call this community our "USH family." While I wasn't born into it, this family has changed me all the same.

This "life-changing" conference is one of many reasons my job doesn't feel like a job. I've been part of all nine of our annual conferences, and my Usher family has grown exponentially. I've observed the change that comes over someone when they finally meet another person with Usher syndrome, someone who 'gets it.' I've felt the energy and excitement that comes from the world's leading experts meeting face-to-face with the families they're working tirelessly to treat. Over the years, attendees from 15 countries and 39 states have come together at these conferences to learn, to share and to join our Usher family. We're thrilled to meet you all in Chicago this year.

Krista Vasi
Executive Director
Usher Syndrome Coalition

USHER SYNDROME COALITION BOARD & STAFF

BOARD OF DIRECTORS

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Father of a daughter with Usher syndrome
President, The Decibels Foundation
Global Director of Information Technology,
L.E.K. Consulting

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Father of a daughter with Usher syndrome
type III
Mediator, Disability Rights Advocate

Margaret Kenna, MD, MPH

Director of Clinical Research
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Communication Enhancement
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Adult with Usher syndrome
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Professor, University of Iowa Carver School
of Medicine

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Adult with Usher syndrome
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Medical School

John Roy

Parent of child with Usher syndrome
Owner, Atlas Scaffolding & Equipment

STAFF

Maria Costa | Director of Development
Julia Dunning, M.Ed. | Events Coordinator
Nancy O'Donnell, MA | Director, USH Trust Director
Krista Vasi, MPA | Executive Director

CONFERENCE INFORMATION

CONFERENCE LOCATION

Chicago Marriott Downtown Magnificent Mile
540 N. Michigan Ave, Chicago, Illinois

The USH Connections Conference will be held in the Marriott Ballroom on the fourth floor of the Chicago Marriott Downtown Magnificent.

A designated guide dog relief area is located on the 9th floor terrace. The doors will be unlocked from 5:30 AM-10:30 PM. If guests need to access the area outside of those hours, dial "0" the door will be opened for you.

CHILDCARE will be provided in the Grace room on the fourth floor.

MEETING ROOM WIFI ACCESS

SSID/NETWORK NAME: Marriott_Conference
Password: usccc2017

USH2017 PLANNING COMMITTEE

Maria Costa
Julia Dunning
Krista Vasi
Lindsey Whyte

INTERNS

Bella Dunning
Emily Losier
Mateo Petracchi
Becca Pickett

The Usher Syndrome Coalition's mission is to raise awareness and accelerate research for the most genetic common cause of combined deafness and blindness. The Coalition also provides information and support to individuals and families affected by Usher syndrome

USH2017 EXHIBITORS

Carroll Center for the Blind

A nonprofit agency, located in Newton, MA, that has served the needs of blind and visually impaired persons of all ages by providing rehabilitation, skills training, and educational opportunities to achieve, independence, self-sufficiency, and self-fulfillment.

Cochlear Americas

Cochlear is the world leader in advanced hearing technologies. For over 30 years, Cochlear has brought the miracle of sound to more than 350,000 people worldwide with its cochlear implant and bone conduction solutions. To learn more about these life-changing technologies, visit Cochlear.com/US.

Foundation Fighting Blindness

FFB is the world's largest funder of retinal degenerative research, and has raised more than \$750 million to fund grants. The urgent mission of the Foundation Fighting Blindness is to drive the research that will provide preventions, treatments, and cures for people affected by retinitis pigmentosa, macular degeneration, Usher syndrome, and the entire spectrum of retinal degenerative diseases.

Hear See Hope

Their mission is to support Usher syndrome research and awareness. Through excellent focus, they can create, communicate and gain knowledge of this currently incurable retinal disorder. Hear See Hope's funds will be directly focused to Usher syndrome research; and, by doing so, they can target the needs of researchers and scientists. With Hear See Hope's help a cure can be found.

Leader Dogs for the Blind

Founded by three Detroit-area Lions Clubs members in 1939, Leader Dogs for the Blind empowers people who are blind, visually impaired or Deaf-Blind with skills for a lifetime of independent travel, opening doors that may seem to have closed with the loss of sight.

Thanks to the generosity of their dedicated supporters, all programs are provided free to clients, including meals and housing during training, travel and equipment.

Leader Dog programs are crafted to address individual situations and adapt to their clients' changing needs at any point in their lives. From youth camp to orientation and mobility cane training, through guide dog training and GPS technology integration, Leader Dogs' programs give clients the confidence and skills they need to live independent lives. Leader Dogs is recognized as a "Best In America" Charity by the Independent Charities of America (ICA).

MED-EL

Founded by world-class scientists and engineers to provide innovative solutions for those dealing with hearing loss. Through revolutionary hearing-implant technology, MED-EL connects people in every walk of life to the rich arena of sound.

USH2017 EXHIBITORS

Second Sight Medical Products

Second Sight is the developer and manufacturer of the Argus II® Retinal Prosthesis System -- the first and only approved long-term therapy for people living with blindness due to retinal degenerative diseases, such as Retinitis Pigmentosa, in the U.S. Argus II is designed to restore some level of vision to people who are profoundly blind and provide them with increases in orientation and mobility skills and is intended to increase their independence and quality of life by restoring their ability to see objects and motion.

Sorenson Communications, LLC

Sorenson is the leading provider of Video Relay Service (VRS), which connects Deaf people who use sign language with the hearing world. Sorenson's innovative products and services offer Deaf callers the option to conduct real-time calls (including 911 emergency calls) through a qualified American Sign Language (ASL) interpreter.

Sprint Accessibility

Sprint Accessibility has specialized programs and services for Deafblind individuals in USA. One most important component of Sprint Accessibility is Sprint IP Relay that is fully accessible to DeafBlind individuals who use Large Print and Braille formats to have live telecommunications access 24/7/365. Sprint IP Relay is a free service to DeafBlind persons. Deafblind persons can use Sprint IP Relay on Internet and Sprint IP Relay Mobile on their Androids and iPhones.

Usher Syndrome Coalition

The Coalition is working to raise awareness and accelerate research, while providing information and support to impacted individuals and families. We strive to be the most comprehensive resource for the Usher syndrome community, bridging the gap between researchers and families. Learn more: www.usher-syndrome.org.

Walk in My Shoes

The writers of this unique collection of 27 powerful stories offer a glimpse into living with Usher syndrome, a progressive disease leading to blindness and deafness. All proceeds from book sales will be donated to the [Usher Syndrome Coalition](http://www.usher-syndrome.org) to help fund scholarships to its [annual conferences](#) and to support research for a cure.

MEET THE SPEAKERS

Edwin Stone, MD, PhD, Keynote Speaker



Dr. Edwin M. Stone is the Director of the Stephen A. Wynn Institute for Vision Research at the University of Iowa. He is well known for his work in defining the genetic basis of blinding eye diseases: ranging from two of the most common causes of blindness, macular degeneration and glaucoma, to much rarer conditions like retinitis pigmentosa and Usher syndrome. Dr. Stone has been very active in removing the technical, legal and financial barriers between genetic discoveries and the patients who could benefit from them.

He founded the Carver Nonprofit Genetic Testing Laboratory at the University of Iowa that provides low cost genetic tests for more than 20 different inherited eye diseases to patients in more than 60 countries. His current interest is in developing affordable gene- and stem-cell-based treatments for all molecular forms of inherited retinal disease.

Dr. Stone received his MD and PhD from the Baylor College of Medicine and his training in ophthalmology and vitreoretinal surgery at the University of Iowa where he joined the faculty in 1990. He holds the Seamans-Hauser Chair of Molecular Ophthalmology in the University of Iowa Carver College of Medicine.

Bill Barkeley, Featured Usher Syndrome Speaker



Bill Barkeley is a deaf-blind adventurer, storyteller and public speaker who speaks with people around the world - those with physical challenges or not - on building a pioneering, adventuring spirit and overcoming the challenges in their lives. Bill's work is about helping others get to a better place in this world and paying it forward in a life that has been rich beyond his wildest imagination.

Mark Dunning, Founder and Chairman, Usher Syndrome Coalition



Mark Dunning is the father of an 18-year-old daughter with Usher syndrome, founding member and Chairman of the Usher Syndrome Coalition, and co-founder of the Decibels Foundation. Mark is also the Global Director of Information Technology for L.E.K. Consulting.

MEET THE SPEAKERS

Ian Han, MD



Dr. Han is Assistant Professor of Ophthalmology in the Wynn Institute for Vision Research at the University of Iowa, Carver College of Medicine. He received his MD from the Duke University School of Medicine, where he was a Howard Hughes Medical Institute research fellow. He completed ophthalmology residency and vitreoretinal fellowship at the Wilmer Eye Institute, Johns Hopkins Hospital, where he also served as Assistant Chief of Service. Dr. Han's research involves translational models of gene and stem cell therapy for inherited eye disease, as well as advanced retinal imaging for improved understanding of vitreoretinal and choroidal pathology.

Michelle L. Hastings, Ph.D.



Dr. Michelle Hastings is an Associate Professor of Cell Biology and Anatomy at The Chicago Medical School at Rosalind Franklin University of Medicine and Science. Dr. Hastings earned an undergraduate degree in biology from St. Olaf College and went on to receive a Ph.D. in biology from Marquette University. She was a postdoctoral and senior fellow at Cold Spring Harbor Laboratory before she joined the faculty at the Chicago Medical School in 2007. She studies the molecular mechanisms of Usher syndrome and therapeutic approaches to treat the disorder using antisense technology.

Her work describing an antisense molecule that rescues hearing and vestibular dysfunction in Usher type 1C was one of the first studies to demonstrate the feasibility of recovering hearing and vestibular function in an animal model of a human disease involving congenital neurosensory hearing loss.

Ben Shaberman



For more than 12 years, Ben has been writing stories on science and research for all of FFB's publications including its Web site, blog, and newsletters. He also presents the latest advancements in retinal research at events and staff meetings. His responsibilities include helping donors one-on-one to understand their conditions, and the steps they can take to manage their vision and potentially gain access to future treatments. Loyola University Maryland published Ben's short story collection titled "Jerry's Vegan Women" in October 2015. Ben's

book of essays, "The Vegan Monologues," was published by Loyola in 2009. His freelance essays and commentaries have been carried by the Washington Post, Chicago Tribune, National Public Radio, and a variety of other newspapers, magazines, and literary journals. Ben holds a master of arts in writing from Johns Hopkins University, a master of science in systems management from the University of Maryland, and a bachelor of science in computer information science from Cleveland State University.

MEET THE USH PANEL

Lynne Murphy Breen



Lynne is a senior underwriting attorney for Chicago Title and Commonwealth Land Title Insurance Companies, part of Fidelity National Financial. She and her husband Bob have two daughters, Harper, 5, and Tatum, 3. They live, along with their two dogs, in North Reading, Massachusetts.

Harper did not pass the newborn hearing screening at birth. After two follow up tests at Children's Hospital Boston, she was diagnosed with bilateral sensorineural moderate hearing loss. She received hearing aids soon thereafter, and began early intervention services including speech therapy, teacher of the deaf/HoH sessions, and music therapy. She was also fortunate to be able to attend the Decibels sponsored early intervention program at the Minuteman Arc for hearing loss.

At five years old, Harper is well advanced in both receptive and spoken English language as compared to her typically hearing peers, and excelling in early literacy, and socially in a mainstream school system with supportive services. Harper also maintains close relationships with other children with hearing loss/deafness. On March 14, 2013, as a result of genetic testing, Harper was formerly diagnosed with Usher Syndrome 2A.

As a mother of a young child with Usher, Lynne is hopeful that treatments and a cure will be available before her daughter has significant vision loss. She continues to be inspired by the many men and women that she has met in the Usher community and their determination to live a full life.

Linnea Haga



Linnea is a 16-year-old from Tampa, Florida, and was diagnosed with Usher Syndrome type 1B in 2014. She was born profoundly deaf and received her first cochlear implant at 18 months old and her second at 11 years old. Her hobbies include reading, art of all kinds and horseback riding. Linnea attends Pasco High School where she will be a senior in the fall. She is currently in an academically rigorous program known as the Cambridge program in order to prepare her for college. Her future plans consist of attending a university - most likely University of South Florida - and pursuing a degree in either animation or medical sciences. Linnea frequently speaks about Usher syndrome and the importance of self-advocacy in regards to being deaf-blind, and has organized an Usher Syndrome Awareness event at her school. She has traveled to many events in Florida and other states in her quest to educate, and has been featured in a Pasco County school board video that has been shared thousands of times on Facebook. She continues to travel and speak publicly about her condition to bring awareness to Usher syndrome.

MEET THE USH PANEL

Derrick Phillips



Derrick Phillips, a Chicago native, was diagnosed with Usher syndrome type 2 as a teenager. Currently, he is superintendent at the Illinois Center for Rehabilitation and Education, a position he's held for over a decade. He is an ordained minister who holds a B.A. in Business Administration and a Master's in Counseling and Guidance from Roosevelt University.

Derrick is a member of the Illinois Deaf Blind Advisory Board. He completed four marathons, published two books, and received numerous awards for motivational speaking. His future plan is to become a national motivational speaker with a message of encouragement especially for those with disabilities.

Kevin Richmond



Kevin identifies as a DeafBlind, gay, Vermonter. He is a professor of American Sign Language and Understanding Deaf Culture courses at the University of Vermont. Formerly a foster father, he proudly adopted his Deaf son. At age 15, Kevin was diagnosed with Usher syndrome, Type 1B. In years 2014 and 2015, he visited Seattle and found his DeafBlind community. This experience helped Kevin accept who he was. Kevin uses ProTactile for communication. He thinks it's important that

DeafBlind folks know that they're not alone and for the Deaf community to support us as a community. Hopefully this will help others accept who they are.

Diana Velarde Torres



Diana was diagnosed 8 years ago with Usher Syndrome type 2 (still to be confirmed genetically). Born and raised in Mexico, Diana has a Bachelor in Science in Electronic Systems Engineering from ITESM (Mexico), and a Master of Business Administration from Newcastle University (UK). After living in Mexico, Canada and the UK, today she lives in Mexico City, where she works for a top professional services firm as a Deals Advisory Manager. Diana is a passionate reader, traveler, and one

of the writers of the book "Walk in my Shoes".

Moira M. Shea (Moderator)



Moira Shea has Usher syndrome 2A. She served as vice chair for the Usher Syndrome Coalition for the past four years. Moira has been involved in the fight to eradicate Usher syndrome for over 40 years.

She served on the Board of Directors for the Foundation Fighting Blindness for 35 years. Moira retired as a federal executive from the US government, which includes 5 years on Capitol Hill. Moira holds a Master in Public Administration from Harvard University's Kennedy School of

Government. She resides in Washington, D.C. with her husband Christophe Lorrain, guide dog, Finnegan and his partner golden retriever, Asia.

RESEARCH PRESENTATION ABSTRACTS

Curing Usher Syndrome

Edwin M. Stone, M.D., Ph.D. – Keynote Speaker

The Stephen A. Wynn Institute for Vision Research
The University of Iowa Carver College of Medicine

There are dozens of single-gene disorders that affect the structure and function of the outer retina. The most common of them affects only one in ten thousand people while the rarest ones affect fewer than 50 people in the United States. We need to be able to treat all of these patients, regardless of how rare or how advanced their disease is. And if we are going to sustainably deliver these treatments to the tens of thousands of people who need them, we will need to be able to do it for less than fifty thousand dollars per patient. This is because without some type of very significant governmental or actuarial wealth redistribution (i.e., more taxes or more extensive insurance coverage), the vast majority of people in the US will not be able to afford treatments at the one-million-dollar price point that many commercial entities are proposing. Although Usher syndrome is the most common cause of combined deafness and blindness in the developed world, some of the associated genes are extremely rare and cause disease in only a few dozen people in the country. Also, some Usher patients have such advanced disease that they will need new photoreceptor precursor cells transplanted under their retina to regain some of their vision. This presentation will present an overview of the science and the strategy behind a nonprofit effort to develop affordable treatments for all genetic types and stages of Usher syndrome.

Gene and Stem Cell Therapy for Usher Syndrome

Ian C. Han, M.D.

Stephen A. Wynn Institute for Vision Research
The University of Iowa Carver College of Medicine

Vision loss in Usher syndrome occurs due to deterioration of the light-sensing photoreceptor cells at the back of the eye. Two main strategies under investigation for restoring vision in Usher syndrome are gene therapy and stem cell therapy. Gene therapy provides a targeted approach for replacing disease-causing genes. Several forms of gene therapy have already been tested for other causes of inherited blindness, and multiple different methods of gene delivery are currently being developed. Accurate identification of the disease-causing genetic variant is needed to properly target treatment. Also, gene therapy requires viable cells to work, and thus holds the most promise earlier in the disease course. Photoreceptor cells have limited ability to regenerate after they deteriorate. Thus, a strategy to replace lost cells is necessary for more advanced disease. Stem cells are multifunctional cells that can be programmed to form new photoreceptors. Transplantation of stem cell-derived photoreceptors may be used to replace degenerative cells and restore vision. Current technology allows for stem cells to be taken from the patient's own skin, thus avoiding the risks of immune system rejection after transplantation. This presentation will provide an overview of ongoing research in both gene therapy and stem cell therapy for the treatment of vision loss due to Usher syndrome.

RESEARCH PRESENTATION ABSTRACTS

The Use of Antisense Technology for the Treatment of Usher Syndrome

Michelle L. Hastings, PhD

Chicago Medical School, Rosalind Franklin University of Medicine and Science,

Drug discovery for the treatment and prevention of Usher syndrome has largely focused on small molecule drugs and gene-delivery approaches. We are developing a novel therapeutic approach using antisense technology to treat Usher syndrome. We have demonstrated that an antisense molecule that corrects defective gene expression in Usher syndrome type 1C (Ush1C) effectively prevents the hearing loss and vestibular deficits associated with the disorder in mice that are genetically modified to carry the causative human Ush1C mutation. Remarkably, a single treatment with the antisense molecule resulted in long-term improvements in hearing and spatiotemporal behavior associated with balance in these mice. Furthermore, delivery of the antisense drug to the amniotic cavity of mice carrying Usher mouse embryos resulted in early correction of *Ush1c* gene expression that persists for up to a month after birth, demonstrating the utility of using antisense technology for the early *in utero* treatment of Usher syndrome. Delivery of the antisense molecule directly into the eye of Usher mice also partially restores *Ush1c* levels in the retina and improves photoreceptor structure and function. Our successful treatment of Usher syndrome in an animal model of the disease is a striking demonstration of the potential for this therapeutic as a treatment for this form of Usher syndrome, and also suggests the potential of this type of therapeutic in the treatment of other forms of visual, hearing and vestibular dysfunction.

¹This work was supported by the NIH/NIDCD, Hearing Health Foundation, Foundation Fighting Blindness, Midwest Eye Banks, Capita Foundation and the National Organization for Hearing Research

² Michelle Hastings receives funding from Ionis Pharmaceuticals.

FFB-CRI's RUSH2A Study: Gaining Insights into USH2A

Ben Shaberman

Foundation Fighting Blindness

Mutations in the USH2A gene are the leading cause of Usher syndrome type 2 and the most common cause of autosomal recessive retinitis pigmentosa (arRP) in the United States. The Foundation Fighting Blindness Clinical Research Institute (FFB-CRI) has launched a four-year, multi-center natural history study of 120 people with USH2A mutations. The study — known as RUSH2A (“R” stands for “rate of progression”) — is taking place at 20 clinical sites around the world. The goals of the study include: 1) Identifying a precise and sensitive outcome measure that can be used in clinical trials for emerging USH2A therapies, 2) Openly sharing knowledge and data gleaned from the study with researchers and companies to facilitate and accelerate therapy development, and 3) Identifying potential participants for future clinical trials. The RUSH2A study is enrolling people genetically diagnosed with USH2A (two mutations) causing Usher syndrome type 2A or arRP. Ben Shaberman’s presentation will include discussion of: the study’s goals, participant criteria, outcome measures, and participant recruitment.

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