

## 7<sup>th</sup> Annual Usher Syndrome Family Conference #USH2015

Saturday, July 11, 2015 | New Orleans, Louisiana

## **PROGRAM**



SCHEDULE AT A GLANCE
Saturday, July 11, 2015

Continental Breakfast and Registration
Blain Kern Ballroom, First Floor
Welcome Address
Former Louisiana Governor Kathleen Babineaux Blanco
Protecting and Restoring Sight: How Novel
Molecular Principles Can Be Harnessed
Nicolas G. Bazan, MD, PhD
Staving Alive: Saving the Retina through
Neuroprotection
Ben Shaberman
Break/Networking
Advances in Drug Therapy for Usher
Syndrome
Iennifer Lentz PhD
Vision for the Euture: Stom Coll Therapies for
Eve Disesses
Dennis Ciegg, PhD
Lunch/Networking
An Update on the UshStat Gene Therapy Trial
for Usher Syndrome Type 1B
Mark Pennesi, MD, PhD
Open Research Q&A
Moderator: Jennifer Lenz, PhD
Break/Networking
Family Panel Discussion
Moderator: Moira Shea
Speakers: Annie Arabie, Dan Arabie, Carol Brill,
Jessica Chaikof, Conner McKittrick
Closing
Mark Dunning
Evening Social at Mulate's Party Hall
743 Convention Center Blvd. New Orleans
Food and fun with our USH family

## WELCOME

Welcome to the Seventh Annual Usher Syndrome Family Conference. This is one of my favorite days of the year. It's not only a way to learn about the latest on the management and treatment of Usher syndrome, it's also an incredible opportunity to network with other families and some of the leading Usher syndrome researchers in the world.

The USH2015 program includes presentations on the progress of neuroprotection, gene therapy, drug therapy, and stem cell therapy to treat Usher syndrome, and the role of the Usher syndrome community in the search for a cure.

The family panel discussion, research Q&A session, lengthy breaks and evening social allow you time to connect with others impacted by Usher syndrome and researchers. We also have activities planned for kids of all ages. It's a unique opportunity for kids with Usher to be the majority for once.

## Mark Dunning

Chairman, Usher Syndrome Coalition

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



## USHER SYNDROME COALITION BOARD & STAFF

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Parent of child with Usher Syndrome President, The Decibels Foundation Director of Information Technology, L.E.K. Consulting

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Adult with Usher Syndrome U.S. Department of Health and Human Services (retired)

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

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#### INTERNS

Rose Borg Kelly Connors Samantha Lozada Priya Sharma

#### STAFF

Julia Dunning, MEd Krista Vasi, MPA

## USH2015 FAMILY CONFERENCE INFORMATION

#### **CONFERENCE LOCATION**

New Orleans Downtown Marriott at the Convention Center 859 Convention Center Boulevard New Orleans, LA 70130

The conference will held on the first floor in the Blaine Kern Ballroom. **CHILDCARE** will be provided in the Tchoupitoulas Room on the second floor.

#### WIRELESS INTERNET ACCESS AT THE CONFERENCE

Complimentary WiFi access will be provided in the conference area.

#### Network: Marriott\_Conf

Password: USH2015 (not case sensitive)

#### DOWNLOAD THE #USH2015 MOBILE APP

Conference details can be found on our mobile app. Download the app on your iPhone, iPad or Android. Visit <u>https://crowd.cc/s/69Xu</u> from your mobile device, or search Usher Syndrome Coalition in the app store.

#### **USH2015 PLANNING COMMITTEE**

Mark Dunning Julia Dunning Elise B. Faucheaux Jennifer Lentz, PhD Moira M. Shea Krista Vasi



### **2015 FAMILY CONFERENCE EXHIBITORS**





#### Affiliated Blind of Louisiana MEDSEL HUMAN DEVELOPMENT CENTER BUILDING CAPACITY-INSPIRING CHANGE Hear now. And always MEDSEL DECOMPOSITION OF A CONTROL OF A CONT

#### Affiliated Blind of Louisiana (ABL) | iCanConnect

Equipment National Deaf-Blind iCanConnect. the Distribution Program (NDBEDP), provides free access to distance communication technologies to people with significant combined hearing and vision loss who meet federal income guidelines. ABL is also a consumer organization which promotes the general welfare of the visually impaired, blind and deaf-blind, educates the public about blindness, and informs consumers of services available visit them. To to learn more. www.icanconnect.org/louisiana.



#### **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 www.Cochlear.com/US.

## Human Development Center | Louisiana Deafblind for Children and Youth

The Louisiana Deafblind Project for Children and Youth (LA-DBP) provides information, services and resources to families and professionals who support children and youth with deafblindness statewide, at no cost. The LA-DBP is funded by the Office of Special Education Programs within the U.S. Department of Education and is administered through the Human Development Center located within the L.S.U. Health Sciences Center in New Orleans, LA. To learn more, visit www.hdc.lsuhsc.edu/ladbp/.

#### Louisiana Relay

Louisiana Relay provides telephone relay service, making it possible for individuals who are deaf, hard of hearing, deaf-blind, or have difficulty speaking to communicate over the telephone.

#### MED-EL

Founded by industry-leading scientists and engineers, MED-EL provides innovative solutions for those dealing with hearing loss. By advancing the field of hearing implant technology, MED-EL's people and products

USHER SYNDR ME COALITION connect individuals around the globe to the rich world of sound. For more information, visit <u>www.medel.com</u> or call 888-MED-EL-CI (633-3524).

## 2015 FAMILY CONFERENCE SPONSORS

#### **GOLD SPONSORS**



#### **The Decibels Foundation**

Founded in 2002, by two families of children with hearing loss, the Decibels Foundation's mission is to provide specialized early intervention, educational services, family support, and access to essential technologies for children with hearing loss from infancy through high school. The programs we fund support children from birth through the time that enter mainstream educational thev а environment and beyond. More importantly, we help the children by first helping their parents learn how to raise a child with a hearing loss, then helping school systems understand what it takes to educate a child with a hearing loss.



#### **Hear See Hope**

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

#### The Megan Foundation

The Megan Foundation raises awareness on Usher syndrome through fundraising events, local and national media coverage, and social media. They also support Usher syndrome research projects that examine potential treatments, and are currently working on support programs for those living with the disorder.



#### Erroll and Suzanne Babineaux Henry Dauterive Moira Shea and Christophe Lorrain





Hear now. And always



#### **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.

#### **Usher 1F Collaborative**

Usher 1F Collaborative is a 501c3 nonprofit foundation whose mission is to fund medical research to find an effective treatment to save or restore the vision of those with Usher Syndrome type 1F.

#### **Usher 2020 Foundation**

Usher 2020 Foundation is a 501c3 nonprofit dedicated to stopping or slowing the degeneration of vision caused by Usher Syndrome. Several promising therapies are currently being developed to help those with retinal diseases. At Usher 2020, we believe that funding this research now will lead to sight-saving therapies by the year 2020.

#### **USH FAMILY AND FRIENDS SPONSORS**

David Alexander Charlie and Emily Babineaux Elise and Blair Faucheaux Akina Hoshino





## MEET THE SPEAKERS



**Governor Kathleen Babineaux Blanco** "Welcome address"

Kathleen Babineaux Blanco, the first and only woman to serve as Governor of Louisiana, was elected to four different offices during her 24 trailblazing years in public service and never lost an election.

She was the first woman from Lafayette elected to the Louisiana House of Representatives, first woman elected to the Public Service Commission, where she was named chairman, and she was elected Lieutenant Governor for two terms. In the 2003 campaign for Governor, she beat 16 men, many of them prominent political figures.

Governor Blanco served during the rough period after Hurricanes Katrina and Rita devastated coastal Louisiana. She secured over \$29 Billion in Washington, D.C. for Louisiana's recovery effort, over-coming extraordinary early resistance. The investment strengthened levees, rebuilt public infrastructure and awarded housing grants to more than 128,000 families to rebuild their homes.

Despite the intense recovery work, Governor Blanco prioritized education funding and oversaw important education improvements. She won national recognition for successes in economic development and left office having built a solid foundation for recovery, leaving nearly \$2 billion in surplus funds.





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#### Nicolas G. Bazan, MD, PhD Boyd Professor and Director Neuroscience Center of Excellence School of Medicine LSU Health New Orleans

"Protecting and Restoring Sight: How Novel Molecular Principles Can Be Harnessed"

obtained his MD in Argentina and was Dr. Bazan а University College fellow at Columbia postdoctoral of Physicians and Surgeons in New York and Harvard Medical School. He was appointed faculty at age 26 at the University of Toronto, where he conducted seminal studies on responses of the nervous system to injury. He then established a Research Institute in Argentina. In 1981, Dr. Bazan joined the faculty of the LSUHSC, where he became the founding Villere Chair and later established and now heads the Neuroscience Center of Excellence.

Dr. Bazan devoted his life to study fundamental cellular and molecular events underlying retinal degeneration as well as neurodegenerative diseases such as Alzheimer's diseases. Among his awards and recognitions include being elected to the Royal Academy of Medicine, Spain (1996); elected fellow of the Royal College of Physicians of Ireland, Dublin (1999); Endre A. Balazs Prize, International Society of Eye Research (2000); the Proctor Medal, ARVO (2007); the Excellence Award, Annual European Association for Vision and Eye Research, Nice, France (2013); and Chairman of the Board of Governors for the ARVO Foundation (2011-2014). He authored the novels Una Vida: A Fable of Music and the Mind, which was produced as a feature film, and The Dark Madonna: A Fable of Resiliency and Imagination. Both novels explore a better understanding of the deep beauty and complexity of human experience.



#### Dennis Clegg, PhD Professor and Chair University of California, Santa Barbara "Vision for the Future: Stem Cell Therapies for Eye Diseases"

Dr. Clegg earned his BS degree in biochemistry at UC Davis and his PhD in

biochemistry at UC Berkeley, where he used emerging methods in recombinant DNA study to the sensory transduction systems of bacteria. As a Jane Coffin Childs Postdoctoral Scholar at UCSF, he studied neural development and regeneration. He has continued this avenue of research since joining the UCSB faculty, with studies of extra-cellular matrix and integrin function in the developing eye. His current emphasis is in stem cell research, with a focus on developing therapies for ocular disease. Dr. Clegg is the recipient of the UCSB Distinguished Teaching Award in the Physical Sciences, the UCSB Community Affairs Board Award, the National Eye Institute Audacious Goals award, and served as Chair of the Department of Molecular, Cellular and Developmental Biology from 2004-2009. He has been a Frontiers of Vision Research Lecturer at the National Eye Institute, a Keynote Lecturer at the Stem Cells World Congress, and a TEDx speaker. He is founder and Co-Director of the UCSB Center for Stem Cell Biology and Engineering, and serves on advisory boards for

the California Institute for Regenerative Medicine and the National Institutes of Health Center for Regenerative Medicine. He is a Co-Principal Investigator of The California Project to Cure Blindness, a multi-disciplinary effort to develop a stem cell therapy for Age-Related Macular Degeneration.



#### Mark Dunning Chairman Usher Syndrome Coalition

Mark Dunning is the father of a sixteenyear-old daughter with Usher syndrome, founding member and Chairman of the Usher Syndrome Coalition, and co-founder and President of the Decibels Foundation.



**USHER SYNDR** ME

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#### Jennifer Lentz, PhD Assistant Professor LSU Health New Orleans

"Advances in Drug Therapy for Usher Syndrome"

Dr. Jennifer J. Lentz is an Assistant Professor of Otolaryngology & Biocommunications, an adjunct faculty

member of the Department of Ophthalmology and a member of the Neuroscience Center of Excellence at Louisiana State University Health Science Center in New Orleans, Louisiana (LSUHSC-NO). The overall goal of her research is to develop a therapeutic approach to prevent or cure the deafness and blindness associated with Usher syndrome (Usher), the most common genetic cause of combined deafness and blindness. Approximately 6-8% of all type 1 Usher cases are caused by mutations in the USH1C gene, which encodes the protein harmonin. The c.216G>A mutation in USH1C accounts for nearly all Usher 1 cases in Acadian populations of south Louisiana, USA and Quebec, Canada. Her laboratory created a model of USH1C by knocking-in the 216A mutation responsible for Usher in an Acadian patient of south Louisiana. The Usher mice have profound deafness, vestibular and retinal dysfunction similar of human Usher. This Usher mouse model and other cellular models are used to understand the underlying mechanisms that lead to the dual sensory loss associated with Usher syndrome, and to develop therapies aimed at preventing or curing deafness and blindness.



Mark Pennesi, MD, PhD Assistant Professor, Opthalmology Oregon Health and Science University "An Update on the UshStat Gene Therapy Trial for Usher Syndrome Type 1B"

Dr. Pennesi studied in bioengineering at the University of Pennsylvania and graduated

Summa Cum Laude. He earned a combined MD/PhD from Baylor College of Medicine in Houston, TX. His PhD degree focused on retinal electrophysiology and animal models of retinal degeneration in the laboratory of Dr. Samuel Wu. He spent his internship at Scripps Mercy Hospital in San Diego and subsequently received his residency training in Ophthalmology at the University of California-San Francisco. Under the mentorship of Dr. Richard Weleber, he earned a fellowship in Ophthalmic Genetics, Electrophysiology, and Medical Retina. He is currently an Assistant Professor in the Department of Ophthalmology at Casey Eye Institute. His clinical and research interests include understanding inherited diseases of retinal degeneration as well as developing novel therapies for these diseases. His interests include retinal imaging using adaptive optics and hand-held spectral domain OCT.



#### Ben Shaberman Director, Science Communications Foundation Fighting Blindness (FFB) "Staying Alive: Saving the Retina through Neuroprotection"

For more than a decade, 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.

Ben's book of essays, "The Vegan Monologues," was published by Loyola University (Maryland) in 2009. The Washington Post, Chicago Tribune, National Public Radio, and a variety of other newspapers, magazines, and literary journals have carried his freelance essays and commentaries. Later this year, Loyola will be publishing Ben's short story collection titled "Jerry's Vegan Women."

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 FAMILY PANEL



My name is **Annie Arabie**. I am a second generation deafblind adult, also known as DeafBlind Children of the DeafBlind Adults (DBCODBA). Like my husband Dan, I was born with Usher Syndrome and have two hearing-sighted sons. I am alumni of Louisiana School for the Deaf. There, I graduated, salutatorian, Class of 1990. I'm

currently employed at Marshall's. I also volunteer many hours at the Amelia Manor Nursing Home where my husband Dan is employed. This nursing home has the largest population of deaf and deafblind residents in the state of Louisiana.

Greetings and Cajun Hugs! My name is **Dan Arabie**. I was born with Usher syndrome in Louisiana, which has the highest incidence of Usher syndrome in America. I graduated valedictorian of my class in 1980 from the Louisiana School for the Deaf. After raising four daughters and retiring from the United States Postal Service, I devoted my time to serving the deafblind and deaf communities as a leader and advocate. My studies in psychology at South Louisiana Community College, the University of Louisiana at Lafayette, and Gallaudet University in Washington, D.C. have helped me to better understand the needs of deafblind people everywhere.

I have served as president of the Louisiana Acadiana DeafBlind Citizens, American Association of the DeafBlind and Lafayette Athletic Association of the Deaf. I also served as vice-president of the Louisiana School for the Deaf Alumni Association and of Affiliated Blind of Louisiana, and as general chairman of the National Softball Association of the Deaf and State Treasurer of the Louisiana Association for the Deaf.

One of my greatest achievements was organizing and leading a DeafBlind rally at the State Department of Social Services in Baton Rouge to request Support Service Providers (SSP) Services for the DeafBlind people of Louisiana. This rally was instrumental in helping our state pass new SSP legislation.

I am also the founding Chief Executive Officer of Deaf Bayou e-News, one of the nation's most popular publications geared toward the deaf and deafblind. The website can be found at <u>www.deafbayou.com</u>



**USHER SYNDR** ME

COALITION

**Carol Brill** lives in Dublin, Ireland, and has Usher Syndrome Type 2a. She is a single mother to her daughter, Sara and is a passionate Ambassador for Usher Awareness and her current campaign is to have deafblindness recognised as a unique disability in Ireland. She works very closely with the Irish national organizations,

Fighting Blindness and the Ann Sullivan Foundation. Her book, *Seeing Disney*, launching in 2015, is a story of the race against time whilst living with Usher Syndrome. Her previous roles include National Chairperson of Fighting Blindness, and amongst many voluntary charity commitments, she held management committee memberships of the European Usher Syndrome Network, Retina International and Deafblind International. Her achievements include a Masters degree in Coaching and Mentoring Practice and a Bachelors degree in Honors Psychology.



**Jessica Chaikof** has Usher Type 1F and is a rising sophomore at Wheaton College in Norton, Massachusetts. She is intending to major in chemistry with the hope of going into teaching at the high school or college level. At the age of fifteen months in May of 1996, she received her first cochlear implant and was the youngest child in the

country to be implanted at the time. In May of 2004, Jessica went bilateral and was diagnosed with Usher syndrome about two years later. Despite the Usher diagnosis, Jessica refuses to let anything stop her. She has been to Australia, England, Germany, Poland, and other parts of Europe. Like any other young adult, she loves Harry Potter and Doctor Who. Her hobbies include 3-D art, such as origami and ceramics, reading, traveling, and writing.



**Conner McKittrick** is a 16 year old with Usher Syndrome Type 1B. Born profoundly deaf, he received his first cochlear implant at age 1. At age 5, he was diagnosed with Usher Syndrome and about a year later, received his second cochlear implant. Conner has attended mainstream school from K- 9<sup>th</sup> grade. He'll be attending

Seattle Christian School next year in the 10<sup>th</sup> grade. From Kindergarten through 6<sup>th</sup> grade he attended private school and from 7<sup>th</sup> to 9<sup>th</sup> grade, he was in a small public school. He receives very little services from the school district, an FM system and orientation and mobility training.

Conner enjoys public speaking, reading and just received his black belt in taekwondo. He has 3 brothers: Cole (14), Hunter (8) and Dalton (7). Dalton also has Usher Syndrome. Conner is very active in the Hear See Hope Foundation, the foundation that his parents started a little over 10 years ago.



**Moira M. Shea** has held a number of government posts, including Congressional staffer and as an economist specializing in international trade and technology development. She's been involved with the Foundation Fighting Blindness since 1980, and is currently a member of its board of directors. Moira also serves as Vice Chair

on the Board of Directors for the Usher Syndrome Coalition.



#### Protecting and Restoring Sight: How Novel Molecular Principles Can Be Harnessed Nicolas G. Bazan, MD, PhD

A consequence of increased life expectancy is a rise in the occurrence of photoreceptor and brain neuron survival failure, as reflected by age-related macular degeneration and other neurodegenerative diseases. Why does this happen? What can we learn by teasing out the intimacy of this failure to help us develop effective therapies?

This issue is magnified when looking at early onset retinal degenerations up close, particularly Usher syndrome. Retinal development, as is the case with the rest of the central nervous system, is driven by neuronal apoptotic cell death, and thereafter neurons, including photoreceptor cells, are considered post-mitotic cells. In retinal degenerative diseases, untimely cell death is set in motion, leading to photoreceptor cell loss due to failed biology, not due to developmental failure. There is a beautiful interdependent relationship between photoreceptor cells and retinal pigment epithelial cells (RPE), whereby a tightly-regulated recycling of key molecules takes place driven by the daily shedding of photoreceptor cell tips and RPE phagocytosis. Cone photoreceptor cell retinoid recycling also involves the Müller cell. The omega-3 docosahexaenoic acid (DHA) attains its highest concentrations in the human body in the photoreceptor cells, and it is remarkable that this fatty acid is retained and conserved between photoreceptor cells and RPE. The outer and disk membranes of photoreceptor cells feature phospholipids richly

**USHER SYNDR** ME

COALITION

endowed with DHA acyl chains. Although age is the main risk factor for age-related macular degeneration, not everyone develops this disease during aging. Despite decades of important findings about signaling that sustains functional integrity of photoreceptor cells and RPE, the decisive mechanisms that sustain the survival of these cells remain incompletely understood. Thus, there is a molecular logic that sustains photoreceptor cell survival and the potential significance of transcriptional signatures in RPE directed by DHA-derived lipid mediators.

- Bazan NG. Molecular Principles for Decoding Homeostasis Disruptions in the Retinal Pigment Epithelium: Significance of Lipid Mediators to Retinal Degenerative Diseases. In: *Retinal Degenerative Diseases: Mechanisms and Experimental Therapy*. Springer. 2015 (in press).

- Bazan NG, et al. Decreased content of docosahexaenoate and arachidonate in plasma phospholipids in Usher's syndrome. Biochem Biophys Res Commun. 1986;141:600-604.

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- Calandria JM, et al. NPD1-mediated stereoselective regulation of BIRC3 expression through cREL is decisive for neural cell survival. Cell Death Differ. 2015. doi: 10.1038/cdd.2014.233.

- Rice DS, et al. Adiponectin receptor 1 conserves docosahexaenoic acid and promotes photoreceptor cell survival. Nature Commun. 2015;6:6228.

#### Staying Alive: Saving Retinal Cells through Neuroprotection Ben Shaberman

Researchers are developing several promising approaches for saving and restoring vision for people with Usher syndrome including: correcting genetic defects, replacing lost retinal cells, and implanting artificial retinas. But in some cases, saving vision may simply come down to keeping retinal cells alive, or slowing their degeneration. least Known at as "neuroprotection," this approach isn't just for the retina — it's being developed to preserve and protect several types of neural cells, including brain tissue and cells of the central nervous system.

Ben Shaberman will discuss several potential advantages that neuroprotection holds as a treatment modality, whether used as a primary therapy or in combination with others. His presentation will review various emerging neuroprotective therapies including those based on: small molecules, viral gene delivery, and stem cells.



## *Vision for the Future: Stem Cell Therapies for Eye Diseases* Dennis Clegg, PhD

One promising approach for the treatment of blinding diseases is to develop cellular therapies using ocular cells derived from stem cells. Both human embryonic stem cells and induced pluripotent stem cells (derived from skin) have been shown to give rise to ocular cells, including photoreceptors and retinal pigmented epithelial cells (RPE). Furthermore, scaffolds that support the survival and function of these cells after transplant have been developed for clinical use. Efforts to develop stemcell derived RPE on a scaffold for the treatment of the dry form of age-related macular degeneration are nearing clinical trials, and research to combine RPE cells with photoreceptors is ongoing.

## Advances in Drug Therapy for Usher Syndrome Jennifer J. Lentz, PhD

Treatment options for Usher syndrome currently focus on the rehabilitation of the hearing, balance and vision impairments. New therapeutic approaches to prevent hearing loss or retinal degeneration are actively being investigated both in the clinic and in the research laboratory. Recent advances have been made by targeting the expression of the disease causing genes with small molecule drugs.

Antisense oligonucleotides (ASOs) and translational readthrough small molecules are two new drugs that are being tested in laboratory models of Usher syndrome type 1C (USH1C). ASOs are synthetic strings of nucleic acids that bind to RNA to alter or reduce their ability to make abnormal proteins. Treatment with ASOs of an animal model containing the human USH1C mutation prevents deafness and balance dysfunction.

Our work is currently testing whether ASOs also prevent Usher blindness. Besides ASOs, translational read-through drugs are small molecules that insert an amino acid at the mutation site into the protein when it is being made to prevent premature truncation. Two of these translational read-through drugs have been tested in animal models and are able to restore full-length protein expression. Success with these two new therapeutic approaches targeting two different types of mutations in USH1C holds significant promise for the treatment of USH1C, as well as other forms of Usher syndrome.

#### An Update on the UshStat Gene Therapy Trial for Usher Syndrome Type 1B Pennesi, M.E., Yang, P., McBride, M., Lauer, A.K., Stout, J.T.,Weleber, R.G.

Usher Syndrome Type 1 is a syndromic form of retinitis pigmentosa that results in congenital sensorineural loss, vestibular areflexia, and retinal degeneration. Type 1B Usher syndrome is due to mutations in *MYO7A* and accounts for about 50% of cases of Type I Usher Syndrome. There remains no effective treatment for this disease, but gene replacement therapy has the potential restore a normal copy of *MYO7A* to the affected patients. UshStat<sup>tm</sup>, developed by Oxford Biomedica (now Sanofi-Fovea), is an EIAV based lentiviral vector that carries a normal copy of the *MYO7A* gene. In collaboration with Hopital Quinze Vignt, we have undertaken a phase I/II clinical trial to evaluate the safety of UshStat<sup>tm</sup> in patients with Usher Syndrome Type IB. To date, six patients have been treated without any serious adverse events. The trial is ongoing with a planned enrollment of up to 18 patients.

Funding: Hear See Hope, Foundation Fighting Blindness, Research to Prevent Blindness, Oxford-Biomedica, Sanofi-Fovea



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