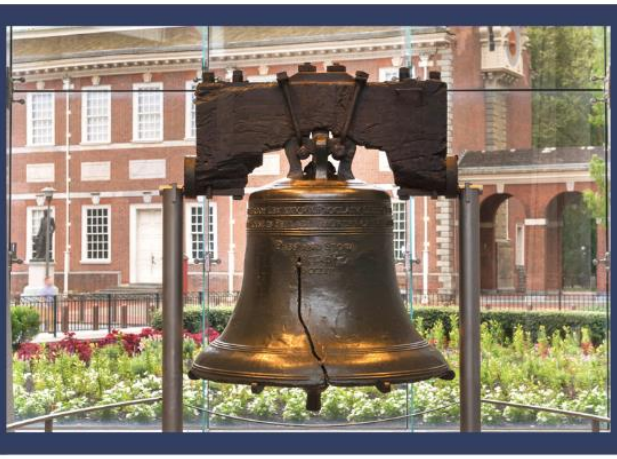
USHER SYNDROME COALITION

USH2019



11th ANNUAL USH CONNECTIONS CONFERENCE

July 13, 2019

PROGRAM

Philadelphia Marriott Downtown | Philadelphia, Pennsylvania

The McKittrick Family has generously funded the USH2019 Family Scholarship Program

Unwavering Commitment

Eloxx Pharmaceuticals is proud to support the Usher Syndrome Coalition

Our Vision: To unlock life's full potential for patients missing essential proteins required for a healthy, normal lifespan.

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NFADB believes that individuals who are deaf-blind are valued members of their communities and should have the same opportunities and choices as others.

NFADB exists to empower the voices of families with individuals who are deaf-blind and advocate for their unique needs.

Stop by our table, and join today!

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Saturday, July 13, 2019
SCHEDULE AT A GLANCE

7:00 – 8:30AM	**Breakfast and Registration:** Level 5 Ballroom, Philadelphia Marriott Downtown
8:30 – 8:45AM	**Welcome Address** Mark Dunning | Chairman, Usher Syndrome Coalition
8:45 – 9:25AM	**"Treatments of the Future for Usher Syndrome: The future is now"** Margaret Kenna, MD, MPH, Boston Children's Hospital | Keynote Speaker
9:25 – 9:45AM	**"Usher Inspiration: From Dreams to Action"** Dario Sorgato | Featured Speaker from the USH Community, Berlin
9:45 – 10:00AM	**"Establishing a translational read-through approach for inherited retinal disorders"** Susan Schneider, MD | Eloxx Pharmaceuticals, USA and Israel
10:00 – 10:30AM	**Break/Networking**
10:30 – 11:00AM	**"RUSH2A Study – the Importance of Natural History Studies"** Christine Kay, MD | Vitreoretinal Associates, Gainesville, Florida
11:00 – 11:30AM	**"Qr-421a, an Antisense Oligonucleotide for the Treatment of Retinitis Pigmentosa Due to USH2A Exon 13 Mutations"** Aniz Girach, MD | ProQR Therapeutics, Netherlands
11:30 – 12:30PM	**USH Research Panel Q&A**
12:30 – 2:00PM	**Lunch/Networking**
2:00 – 2:30PM	**"Importance of Collaboration in the Educational System: Identifying Your Team Players"** Lanya McKittrick, PhD & Carly Fredericks | Parents of kids with Usher syndrome
2:30 – 3:00PM	**"The Benefits of Federal Employment for People with Usher Syndrome"** Ryan Thomason | Adult with Usher Syndrome
3:00 – 3:30PM	**Break/Networking**
3:30 – 4:45PM	**USH Panel Discussion**
4:45 – 5:00PM	**Closing**

WELCOME

Welcome to the 11th annual USH Connections Conference! This will be the largest gathering of people with Usher syndrome and their families in history with nearly 400 people in attendance. Today you are not alone.

You will get to hear about research from some of the world's leading Usher syndrome researchers. You will get to hear about how Usher syndrome figures in to the goals of some pharma companies. And you will get to hear from inspirational members of our community.

Most importantly you will get to meet and connect with others who are facing the same fears and challenges that you are. You will get to share your story and learn theirs. And hopefully like so many of the attendees at this conference, you will make lifelong friendships with people who understand you better than anyone.

I am the chairman and one of the founders of the Usher Syndrome Coalition. I am the father of a twenty year old daughter who cannot attend because she is leading caravans of tourists on trail rides in the Rockies (the girl with poor balance, vision, and hearing has chosen the cowboy life). I look forward to seeing so many of my dearest friends here in Philadelphia. And if we have not met yet, I look forward to welcoming you to the Usher syndrome family. I hope you, like me, come to look forward to this event every year.

Thanks for coming!

Mark Dunning
Chairman
Usher Syndrome Coalition

USHER SYNDROME COALITION
BOARD & STAFF

BOARD OF DIRECTORS

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

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

Margaret Kenna, MD, MPH
Director of Clinical Research
Dept. of Otolaryngology and Communication Enhancement
Boston Children's Hospital

Nancy Corderman
Mother of four, two with Usher Syndrome
Co-founder of the Usher Syndrome Society

Lanya McKittrick, PhD
Mother of four boys, two with Usher syndrome
Co-founder of Hear See Hope Foundation
Special Education Researcher

Stephen Perreault
(Retired) Anne Sullivan Macy Medal recipient for building programs for children who are blind and deafblind in Latin America

Monte Westerfield, PhD
Director, Usher syndrome research laboratory, Institute of Neuroscience,
Professor of Biology, University of Oregon

William Kimberling, PhD
(Retired) Director, Center for the Study and Treatment of Usher Syndrome, Boys Town National Research Hospital
Professor, University of Iowa Carver School of Medicine

Kelley Stidd
Mother of a daughter with Usher syndrome type 2A
Certified Public Accountant

Danay Trest
Adult with Usher syndrome
(Retired) Dept. Business Administrator and Analyst Programmer
University Mississippi Medical Center

Karmen Trzupek, CGC
Genetic Counselor, Informed Medical Decisions (InformedDNA)
Genetic Counseling Team Leader, Ocular

Kevin Richmond
Adult with Usher syndrome
Professor of American Sign Language and Understanding Deaf Culture courses at the University of Vermont

Kathy Thompson, PhD
Adult with Usher Syndrome
Management Consultant

Kadie Trauger
Adult with Usher Syndrome 1B
Former Sign Language Interpreter Coordinator
Stay at home mother of two kids

USHER SYNDROME COALITION
BOARD & STAFF

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Boys Town National Research Hospital

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Research Associate, University of Oregon
Co-Author, Usher Syndrome Blog

Heidi L. Rehm, PhD
Director, Laboratory of Molecular Medicine, Partners Healthcare Personalized Medicine
Associate Professor of Pathology, Harvard Medical School

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

STAFF

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Krista Vasi, MPA | Executive Director

INTERNS

Alaina Bush
Jessica Chaikof
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Karly McNeish

CONFERENCE INFORMATION

CONFERENCE LOCATION

Philadelphia Marriott Downtown
1201 Market Street, Philadelphia, PA 19107

The USH Connections Conference will be held in the Ballroom (Salons C, D and E) on the fifth floor of the Philadelphia Marriott Downtown.

A designated guide dog relief area will be located adjacent to the ballroom. The space will be marked **"Dog Relief Room."**

CHILDCARE for children 9 and under is located in Room 501. The Usher Syndrome Coalition has hired Philadelphia Nanny Network, a highly reputable childcare agency, to provide your child(ren) with a safe, fun and comfortable experience while attendees relax and enjoy the conference.

MINI-CAMP for kids and teens ages 10-17 is located in Meeting Rooms 401 and 402: As an official USH Partner of the Usher Syndrome Coalition, Ava's Voice is pleased to offer USHthis Mini Camp - a new program for youth attending the USH Connections Conference.

PORTRAITS AND STORY SUBMISSIONS

We're excited to host the Usher Syndrome Society as they continue to grow the global "Shine a Light on Usher Syndrome" exhibit featuring portraits and stories of those living with Usher syndrome. This exhibit travels the globe spreading public awareness of Usher syndrome. Portraits will be taken in Meeting Room 502 during breakfast and registration, breaks and lunch. Learn more and sign up here: http://bit.ly/USH2019portraits

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Usher Syndrome Coalition: Connecting the global Usher community. Our mission is to raise awareness and accelerate research for the most genetic common cause of combined deafness and blindness, while providing information and support to individuals and families affected by Usher syndrome.

USH2019 EXHIBITORS

Ava's Voice (USH Partner)
The mission of Ava's Voice is to empower youth with Usher syndrome and to educate families, and school communities.

Eloxx Pharmaceuticals
Eloxx is developing therapies to treat rare premature stop codon diseases. Their development efforts are focused on read through of these stop codons to extend mRNA half-life and enable functional protein synthesis. Their lead drug candidate is in clinical development for cystic fibrosis and cystinosis. They have also initiated an inherited retinal disease program.

Foundation Fighting Blindness
Foundation Fighting Blindness (FFB) is the world's largest private funder of retinal degenerative research, focused on diseases such as macular degeneration, RP and Usher syndrome. They have 50 Chapters throughout the country.

Hear See Hope (USH Partner)
Hear See Hope is a 501c3 nonprofit and a founding partner of the Usher Syndrome Coalition. Their mission is to support Usher syndrome research and awareness efforts.

Helen Keller National Center
HKNC is the only organization of its kind—providing training and resources exclusively to people age 16 and over who have combined vision and hearing loss.

Leader Dogs for the Blind
Leader Dogs for the Blind offers a full-range of services provided free of charge to our clients, including Guide Dog Training, a Deaf Blind guide dog program, Orientation & Mobility Training and Summer Experience Camp.

MED-EL Corporation
MED-EL was 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. For more information, visit www.medel.com or call 888-MED-EL-CI (633-3524).

National Family Association for Deaf-Blind (NFADB)
NFADB is a nonprofit national organization that has served families with individuals who are deaf-blind since 1994. Originally started by and for families, it has expanded to include any interested professionals and other individuals or organizations who wish to empower the voices of families with individuals who are deaf-blind.

New York Deaf-Blind Collaborative (NYDBC)
NYDBC is the federally funded State Deaf-Blind Project for children who are deaf-blind between ages 0 - 21.

USH2019 EXHIBITORS

Sprint Accessibility
Sprint Accessibility provides many products which can assist individuals in connecting the world around them. These services include text relay, and captioned telephones. Whether you are an individual with low vision and some hearing, someone who is low vision and deaf, someone who is hard of hearing and blind, or completely deaf-blind, Sprint Accessibility has tools designed to keep you connected to the world around you.

Usher 1F Collaborative (USH Partner)
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.

MEET THE SPEAKERS

In order of appearance

Margaret Kenna, MD, MPH, Keynote Speaker, Boston Children's Hospital

Margaret Kenna's research focuses on pediatric otology; currently, she is studying the underlying causes of sensorineural hearing loss (SNHL), the most common congenital sensory impairment. Her research includes the genetics of hearing loss, especially GJB2 (Connexin 26) and Usher syndrome; anatomic inner ear anomalies and vestibular function testing; and congenital cytomegalovirus infection. Dr. Kenna's earlier research focused on the causes and treatment of chronic suppurative otitis media (CSOM). She and her colleagues in Pittsburgh established that medical, not surgical therapy was very effective and should be the initial management option.

Dario Sorgato, Featured Speaker from the USH Community

Dario Sorgato was born in Italy in 1978. He grew up in Padua and moved to Milan to study Design. At the age of 16 he was diagnosed with Usher syndrome.

After graduation he undertook several travels, seeking new challenges and determined not to be stopped by sensory limitations. Dario spent one year in Australia and New Zealand, he completed the Camino de Santiago, he joined the crew of the "Research Vessel Heraclitus" and sailed for nearly two years from Cape Town to Havana.

In 2011, he founded "NoisyVision", a non-profit organization, to raise awareness about sensory disabilities and promote activities for people with visual and/or hearing impairments.

Susan Schneider, MD, Eloxx Pharmaceuticals, USA and Israel

Dr. Schneider serves as Senior Vice President of Clinical Development, Ophthalmology at Eloxx. She has nearly 15 years of experience in drug development while serving as Chief Medical Officer at ThromboGenics (now Oxurion), Vice President and Therapeutic Area Head of Retina and Glaucoma at Allergan, Vice President Retina, at Acucela, Sr. Medical Director at GlaxoSmithKline, Director, Clinical and Scientific Affairs for Ophthalmology at Bausch & Lomb, Vice President and Chief Medical Officer, US Clinical Affairs at Santen, and Medical Director at Genentech. While at Genentech, she worked extensively on the ranibizumab program. Her current focus is developing potential therapies for Inherited Retinal Disorders.

Dr. Schneider received her medical degree from the Medical College of Pennsylvania followed by residency in ophthalmology at the Medical College of Virginia. She completed fellowship training in ocular oncology at Thomas Jefferson Wills Eye Hospital and in ophthalmic pathology at The Johns Hopkins Wilmer Eye Institute.

MEET THE SPEAKERS

Christine Kay, MD, Vitreoretinal Associates, Gainesville, Florida

Christine Kay, M.D. is the director of electrophysiology, retinal genetics, and clinical trials at Vitreoretinal Associates in Gainesville, Florida. Prior to this, she was an Assistant Professor and director of the retinal fellowship and retinal genetics service at the University of Florida. She graduated from Harvard University with a magna cum laude in neuroscience, went to medical school at the University of Florida, and completed her vitreoretinal surgical fellowship at the University of Iowa.

After her vitreoretinal surgical fellowship, she was awarded a 5-year Career Development Award from the Foundation Fighting Blindness which funded research focusing on genetic treatment of achromatopsia. She is now a PI in the AGTC-sponsored achromatopsia gene therapy trials. She is also a PI and surgeon in the Nightstar XIRIUS trial for XLRP, and a PI in the Alkeus-sponsored Stargardt disease trial, Foundation Fighting Blindness sponsored RUSH2A trial, and Ophthotech trial for Stargardt disease. She has a large IRD patient population, with over 900 patients in her clinical database and with 650 of these patients genotyped. Dr. Kay was recently inducted into both Macula Society and Retina Society. She has been an invited speaker at the annual Foundation Fighting Blindness VISIONS conference for the past 6 years, is actively involved in FFB's clinical trial consortium for inherited retinal disease, and teaches courses on gene therapy annually at American Society of Retina Specialists and Academy of Ophthalmology.

Aniz Girach, MD, ProQR Therapeutics, Netherlands

Aniz Girach, MD is ProQR's Chief Medical Officer. After having spent 11 years in academia, he joined the pharmaceutical industry with Eli Lilly, focusing on retinal diseases. He has in total 22 years industry experience in roles with Merck, as their Global Head of Ophthalmology, and Alcon, where he was Vice President of Clinical Development, and ThromboGenics (now Oxurion), where he was the Global Head of Ophthalmology/Chief Medical Officer overseeing the development and approval of Ocriplasmin (Jetrea)—a first in class biologic therapy for retinal disease. In addition to an Honorary Professorship at Wills Eye Hospital, Philadelphia, USA, he was recently the Chief Medical Officer at Nightstar Therapeutics, overseeing the development of gene therapies for inherited retinal diseases. He is a member of three Scientific Advisory Boards for international ophthalmic organizations currently, and reviewer for five peer-reviewed journals, including Eye and IOVS. He has edited four books and published over 100 abstracts/manuscripts in peer-reviewed journals in Ophthalmology.

MEET THE SPEAKERS

Lanya McKittrick, PhD, Parent of children with Usher syndrome

Lanya McKittrick received her Ph.D. in Special Education at the University of Northern Colorado. Her dissertation is titled Strategies that Parents of Children who are Deafblind Employ to Foster Collaboration within IEP Teams. Her research interests include: services and supports for students who are deafblind and their families; family-professional partnerships; student-led IEP's; and transition. Her research is rooted in her personal experience as a mom to four sons, two who have Usher Syndrome (Dalton, age 11 and Conner, age 20). Lane co-founded the Hear See Hope Foundation to raise awareness and funding for Usher research and to support families through their Usher journey.

Carly Fredericks, Parent of a child with Usher syndrome

Carly Fredericks has a love for the field of deafblindness. As founder and President of Ava's Voice, she empowers youth with Usher syndrome and educates families and school communities. She is also the Program Coordinator for iCanConnect/NJ, the Deaf-Blind Equipment Distribution Program that provides telecommunications equipment for individuals with combined hearing and vision loss. She has been a Family Specialist for the New Jersey Consortium on Deaf-Blindness and has been on the Board of The National Family Association for the Deaf-Blind (NFADB). Carly is mom to Ava (age 12) who has Usher syndrome (Ava age 12).

Ryan Thomason, Adult with Usher syndrome

Ryan Thomason was born and raised in Lake Stevens, Washington, a small town north of Seattle. Sports was a big part of growing up for him. Ryan loved training and competing in football, basketball and then track and field. After high school, Ryan went to Central Washington University, where he competed on the Track and Field team, got a BA in Administrative Management and Information Technology, and was lucky enough to meet the love of his life, Jennifer. After a nine-year stint in Utah, where both of Ryan's kids (Lincoln and Finley) were born, he enrolled into the MBA program at Weber State University that gave him the tools he needed to start a new career in the Department of Defense as a federal employee in Philadelphia. Ryan and his family have been in Philly since 2016 and have been enjoying everything that the East Coast has to offer for their family!

MEET THE USH PANEL

Jeffrey S. Bohrman, PhD

Dr. Jeffrey Bohrman was born with Usher Type 1C (German Jewish, not French Cajun). He had a strong oral upbringing despite his impaired speech. He did not learn ASL formally until he was 41. After Jeff graduated from Clarke School for the Deaf, he started high school in Philadelphia where his family had settled down after many years growing up in Northeast New Jersey. Jeff went on to graduate high school and earn four college degrees, all without having interpreters, which were not available in those days. His doctorate was in Pharmacology. He worked as a pharmacy intern and became registered in Illinois.

Jeff did his postdoctoral fellowship through the University of Tennessee and worked as a research pharmacologist in a federal agency in Ohio for ten years before being forced to go on disability due to his blindness. After an intensive rehabilitation training, he became an accomplished Program Director of a statewide program for DeafBlind services in Ohio for 21 years. He was an active member and served on the board of local, state, national, and international organizations dealing with DeafBlindness. Jeff has retired to Chicago with his wife of nearly 49 years. They have two children and four grandchildren.

Wasim Raza

Wasim was born in Pakistan; he arrived in America at the age of nineteen, two months after 9/11. Wasim is a graduate of Walden University with a degree in Healthcare Information Systems. Since becoming deaf and blind, Wasim created a series of informative videos under the name, “DeafBlind Warrior”, to help educate people, with and without Usher syndrome. His goal is to bring awareness to Usher syndrome, to people all over the globe.

Sandra Scala

Sandra Scala has Usher syndrome (USH2A), and anxiety, and has been through stages of dark times in her life. She still has her moments; BUT, keeps pushing forward! She is a 39-year-old stay-at-home mom, whose focus in life is her family (three of her own, and four stepchildren, three of which are on the Autism Spectrum, ADHD, and one with Tourette’s Syndrome). She also has a Guide Dog, Mimi.

Sandra has a passion for writing, blogging, and speaking, and is hoping to make a difference through her blog - The Mind, Untangled. She has a book, “New Beginnings” where she shares her thoughts. She was invited to do an Interview on a live television show in Cyrpus, where she shared her story, and this can be found on her website www.theminduntangled.com.

Sandra is at the point where she is ready to share her stories and face her fears. She takes one day at a time, making the most of life and what it has to offer - all in hopes to MOTIVATE and INSPIRE – adults, parents, kids, teens – to keep moving forward, and know that they are NOT alone! “Let’s untangle our mind, and set it free…”

MEET THE USH PANEL

Anne Schueler

Anne Schueler is the mother of soon to be 5 children, ages 12, 9, 7, 3, and 20 weeks gestation. She and her family live in Cincinnati, Ohio with their oldest daughter's service dog, Beyonce. Her daughters Gianna (12) and Pia (7) were diagnosed with USH1B at age 5 and 9 months respectively. Both girls are bilateral cochlear implant users and attend their parish school in a mainstream setting. Since the time of their daughters' diagnosis, she and her husband have committed to finding ways to support them that are practical and positive while also connecting with other families facing the same challenges for guidance and comfort. While watching her daughters deal with this syndrome has been difficult, Anne and her family are privileged to have met so many incredible role models with Usher syndrome and their families, and for that, they are thankful to be a part of the USH family.

Monte Westerfield, PhD, Moderator

Dr. Monte Westerfield is the Director of an Usher syndrome research laboratory and a Professor of Biology at the Institute of Neuroscience, University of Oregon**.** Monte has two adult children with Usher syndrome.

PRESENTATION ABSTRACTS

Treatments of the Future for Usher Syndrome: The Future is Now
Margaret Kenna, MD, MPH – Keynote Speaker
Boston Children's Hospital

Usher syndrome is a genetic deaf-blind syndrome with at least 9 associated genes. Until now, management has been based on supporting the hearing loss with hearing aids, cochlear implants, and language therapy; the vison loss with Vitamin A, antioxidants, and limited exposure to light. Genetic therapy is in its early infancy, but growing. Various techniques targeting different aspects of the genetic code are being developed in the laboratory and in animal models, with many of these techniques potentially applicable to human Usher syndrome. Gene silencing, gene destruction, and gene supplementation are varying approaches to treatment. Additional techniques include gene editing (using CRISPR-Cas9, etc.), replacement genes attached to viral vectors, RNA interference/editing, antisense oligonucleotides, and the use of organoids to study various therapies. Examples of these different treatments include RNA-based therapies for Leber's amaurosis and an exome-skipping strategy for USH2A; CRISPR-based therapeutics for LCA10, eliminating a mutation in the CEP290 gene; the use of stem cells to create functional new cells, and progenitor cell activation, proposed for sudden hearing loss and noise-related hearing loss. Although most of these proposed therapies remain in the lab and experimental, and not all will be applicable to every genetic disease, results are exciting and give hope for the future.

Usher Inspiration: From Dreams to Action
Dario Sorgato
NoisyVision, Italy

It is because of vision and hearing loss that people with Usher syndrome have the possibility to see and hear like no one else. Without denying the problems and hurdles of daily life, people with Usher can learn to understand the signals of their own world and share their uniqueness. The inspiration of what people with Usher can do is not only for the Usher community but also for those who think they see and hear because their eyes and ears are functioning. Given the fact Usher syndrome is a challenging condition, it can be transformed into something positive. Starting from his personal experience and travels through the achievements of the NGO he founded, Dario Sorgato will provide some ideas on how to pursue your goals and think about your next step, perhaps the first, to realize your own dreams.

Establishing a Translational Read-Through Approach for Inherited Retinal Disorders
Susan Schneider, MD
Eloxx Pharmaceuticals, USA and Israel

Purpose: We are developing a novel approach to addressing inherited retinal disorders, including Usher Syndrome, that is being evaluated as a series of Eukaryotic Ribosome Selective Glycosides (ERSGs), delivered to the eye by intravitreal injection. These compounds are optimized to promote read-through of premature stop codons to restore essential proteins that are missing due to nonsense mutations.

Methods: Read-through of Usher mutations, functional restoration of protein production associated with a nonsense mutation, tolerability, and pharmacokinetics of retinal exposure evaluations were performed.

Results: In vitro models demonstrated positive read-through of up to 2.5-fold over baseline with Usher 1F and Usher 2A mutations. Ocular tolerability was assessed in a rabbit model. ERG and histopathology evaluation showed superior tolerability of intravitreally injected Eloxx compounds relative to another read-through inducing agent, gentamicin. There was no adverse effect on intraocular pressure. Substantial retinal exposure was demonstrated.

Conclusion: These results demonstrate positive early evidence of a novel read-through approach for inherited retinal disorders, including Usher Syndrome. The data support continued development of these novel small molecules for intravitreal delivery to restore production of full-length functional proteins in the retina as a potential common therapeutic option for these diseases.

RUSH2A Study: The Importance of Natural History Studies

Christine Kay, MD
Vitreoretinal Associates, Gainesville, Florida

In this presentation, we will begin with a very brief clinical overview of Ush2A-related retinal degeneration, and discuss the both syndromic and nonsyndromic Ush2A. This will be followed by an in-depth discussion of the RUSH2A natural history study design, inclusion/exclusion criteria, outcome measures, and protocol. No RUSH2A results will be released at this time since study and analysis are still ongoing. We will then discuss from a broader perspective the importance and relevance behind natural history studies. Natural history studies are critical in identification and characterization of a patient population, aiding drug development, and informing clinical trial design and outcome measures. We will end with a question and answer session.

QR-421a, an Antisense Oligonucleotide for the Treatment of Retinitis Pigmentosa Due to USH2A Exon 13 Mutations

Aniz Girach, MD
ProQR Therapeutics, The Netherlands

Usher syndrome (USH) is a common inherited retinal degeneration (IRD), which affects hearing, balance and vision. USH caused by USH2A mutations is one of the commonest causes of both USH and RP with syndromic features. Currently there is no treatment for the devastating sensory deficits caused by USH. In an attempt to fill this void, QR-421a is an antisense oligonucleotide (ASO) and designed to specifically target mutations in exon 13 of the USH2A gene for the treatment of patients with RP. QR 421a binds to a specific sequence in the USH2A pre-messenger ribonucleic acid (mRNA) and modulates splicing by enhancing exon-skipping. Skipping of exon 13 from the USH2A pre-mRNA results in mRNA without exon 13, that is expected to lead to functional (albeit shorter) Usherin protein. It is hypothesized that treatment with QR-421a will result in restoration of functional Usherin protein in photoreceptors and restoration of vision. The first-in-human study of QR-421a (PQ-421a-001) has already

commenced and will evaluate the safety and tolerability of a single dose of QR-421a in adult subjects with RP due to mutations in exon 13 of the USH2A gene. QR-421a will be administered via intravitreal injection in the worst eye. A potential dose response relationship and duration of effect following a single dose of QR-421a, based on improvements in retinal structure or visual function, will also be investigated to inform selection of dose level(s) and dosing intervals for subsequent studies of QR-421a clinical development.

Importance of Collaboration in the Educational System: Identifying Your Team Players

Lanya McKittrick, PhD and Carly Fredericks

Hear See Hope, Washington; Ava's Voice, New Jersey (respectively)

Parents of children who have Usher syndrome have unique challenges before, during, and after Individualized Education Program (IEP) meetings. Often there is a lack of professional knowledge about deafblindness, thereby requiring families to gain and share knowledge. Because parent knowledge and advocacy are essential roles, there is a need to understand better how parents advocate and share knowledge during the IEP meeting. The results from this qualitative research study will offer participants an understanding of parent-initiated strategies used to increase IEP team collaboration and to address their child's needs, paying careful attention to the much-neglected area of deafblindness. Families can benefit from hearing from other family's experiences. The leadership skills and strategies identified in this study will provide families with ideas about how to advocate and share knowledge using a firm, but positive approach.

In addition to sharing advocacy strategies identified in the research study, the presenters will share their personal experiences as moms of children with Usher syndrome, offering practical strategies they use when working with educational teams.

The Benefits of Federal Employment for People with Usher Syndrome

Ryan Thomason, Adult with Usher syndrome

Department of Defense, Philadelphia, Pennsylvania

What is the federal mandate to hire people with disables and how can I make it work for me? How a 'Schedule A Letter' will get your name toward the top of the review pile and how to apply for federal jobs. How the Department of Labor's Workforce Recruitment Program benefits current college students and recent graduates with disabilities looking for internships and full-time employment. Learn about how non-profits and businesses use the Ability One program and how this program for the blind is obligated by the federal government to be a first source selection of supply/services contracts

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USHER SYNDROME COALITION

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