

USHER SYNDROME  
COALITION



# 8th Annual Usher Syndrome Family Conference

**#USH2016**

Saturday, July 9, 2016 | Seattle, Washington

**PROGRAM**



# SCHEDULE AT A GLANCE

Saturday, July 9, 2016

8:00 – 8:45AM	<b>Continental Breakfast and Registration</b> Crystal Ballroom, Third Floor
8:45 – 9:00AM	<b>Welcome Address</b> Mark Dunning, Chairman   Krista Vasi, Executive Director
9:00 – 9:30AM	<b>A combined cochlear and vestibular prosthesis for the treatment of Usher syndrome: a clinical trial</b> James Philips, PhD
9:30 – 10:00AM	<b>Hearing and Usher syndrome: diagnosis and management</b> Kathleen C.Y. Sie, MD
10:00 – 10:30AM	<b>Break/Networking</b>
10:30 – 11:00AM	<b>Deconstructing disease genes: How zebrafish models of USH can help develop therapies for vision loss</b> Jennifer Philips, PhD
11:00 – 11:30AM	<b>Unraveling Usher syndrome: The role of genetic testing</b> Karmen Trzupek, MS, CGC
11:30 – 1:00PM	<b>Lunch/Networking</b>
1:00 – 1:30PM	<b>Advances in Stem Cell Research for Treating Retinal Diseases</b> Jennifer Chao, MD, PhD
1:30 – 2:45PM	<b>Research Q&amp;A Session</b>
2:45 – 3:15PM	<b>Break/Networking</b>
3:15 – 4:45PM	<b>Family Panel Discussion</b> Moderator: Moira M. Shea Speakers: David Hebert, Ramona Rice, John Romish, Clare Weigel
4:45 – 5:00PM	<b>Closing</b> Mark Dunning
6:00 – 9:00PM	<b>Evening Social at Rock Bottom Brewery</b> 1333 Fifth Avenue, Seattle

## WELCOME

Welcome to the 8th Annual Usher Syndrome Family Conference. At 200 people, we are sold out for the first time. On the one hand, it saddens us to have to turn away members of the Usher syndrome community. We never want to deny anyone an opportunity to gain knowledge and network with researchers and other families. On the other hand, we should all celebrate this because it is a GREAT problem to have. We are becoming a community that is bursting at the seams, demanding more opportunities, speaking with an ever-larger chorus of voices.

A large active community is a tremendous asset. Community is the only way to combat the social isolation often associated with Usher syndrome. An active community is necessary to push for governmental investment in science. And, as we often note, a large community is critical, absolutely critical, to finding treatments for Usher syndrome. There will be no treatments without clinical trials. There will be no clinical trials without enough Usher syndrome families to participate as candidates. We need everyone to be involved.

Every year we seem to move closer to that goal. So I am both sorry and very, very happy that the family conference sold out.

Next year, in Chicago, we'll be sure to have enough room to welcome everyone. Unless, of course, this community once again exceeds all expectations.

*Mark Dunning*

Chairman, Usher Syndrome Coalition

# USHER SYNDROME COALITION BOARD & STAFF

## BOARD OF DIRECTORS

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Parent of child with Usher syndrome | Chairman, The Decibels Foundation | Director of Information Technology, L.E.K. Consulting

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Adult with Usher syndrome (Retired) U.S. Department of Health and Human Services

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Father of a daughter with Usher syndrome type III  
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Certified Genetic Counselor,  
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### **Jennifer Phillips, PhD**

Research Associate,  
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### **John Roy**

Parent of child with Usher  
syndrome | Owner, Atlas  
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## **STAFF**

Julia Dunning, Events Coordinator  
Nancy O'Donnell, USHTrust Director  
Krista Vasi, Executive Director

# USH2016 FAMILY CONFERENCE INFORMATION

## CONFERENCE LOCATION

Washington Athletic Club in downtown Seattle  
1325 Sixth Avenue, Seattle, WA 98111-1709, USA

The Usher Syndrome Family Conference will be held in the Crystal Ballroom on the third floor of the Washington Athletic Club. **CHILDCARE** will be provided in the Scaylea Room on the third floor.

## USH2016 PLANNING COMMITTEE

Mark Dunning  
Julia Dunning  
Lane McKittrick  
Moirá Shea  
Krista Vasi

## INTERNS

Bella Dunning  
Liam Jones  
Alexandra Lozada  
Samantha Lozada  
Karly McNeish

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**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.

2016 FAMILY CONFERENCE EXHIBITORS

# iCanConnect

The National Deaf-Blind Equipment Distribution Program

# MED EL



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# WASHINGTON RELAY

HKNC



**Helen Keller**  
NATIONAL CENTER  
for Deaf-Blind Youths and Adults

## **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](http://www.Cochlear.com/US).

## **Helen Keller National Center (HKNC)**

Authorized by an Act of Congress in 1967, HKNC provides training and resources to individuals 16 and over who have combined hearing and vision loss. HKNC has a national presence with regional offices in 11 locations. Find your local Regional Representative: [www.helenkeller.org](http://www.helenkeller.org)

## **iCanConnect**

iCanConnect, the National Deaf-Blind Equipment Distribution Program (NDBEDP), provides free access to distance communication technologies to people with significant combined hearing and vision loss who meet federal income guidelines.

## **MED-EL**

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](http://www.medel.com) or call 888-MED-EL-CI (633-3524).



## **Washington Relay Service**

Washington State Office of the Deaf and Hard of Hearing 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.

## **Washington Sensory Disabilities Services**

Washington Sensory Disabilities Services (WSDS) supports the developmental and learning needs of children, ages birth to 21, who are: Deaf/Hard of Hearing, Blind or Visually Impaired, or Deaf-Blind.

## **2016 FAMILY CONFERENCE SPONSORS**

### **GOLD SPONSORS**

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#### **Don & Pam Dunning**

#### **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 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 they enter a mainstream educational 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.



## BRONZE SPONSORS

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### Moira Shea and Christophe Lorrain

#### 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](https://www.cochlear.com/US).



*Hear now. And always*

## 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

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**Caroline Brown and Craig McCaa**

## USH FAMILY SCHOLARSHIP PROGRAM SPONSORS

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### Global Genes

The Coalition was one of several grant recipients selected from a pool of 137 applicants. Global Genes is committed to meeting the collective need of the 350 million patients and hundreds of advocacy groups fighting for treatments and cures for over 7,000 rare diseases.



## MEET THE SPEAKERS

### **James Philips, PhD**



James O. Phillips, Ph.D. joined the faculty at the University of Washington in 1998, and is currently a Research Associate Professor in the Department of Otolaryngology-Head and Neck Surgery. He is also the Director of the Dizziness and Balance Center at the University of Washington Medical Center (UWMC), the Vestibular Diagnostic Laboratory at UWMC, the Roger Johnson Clinical Oculomotor Laboratory in the Division of Ophthalmology at Seattle Children's Hospital. Prior to joining UW, he earned a Ph.D. in Psychology and in Physiology from the University of Washington. Dr. Phillips teaches in the Departments of Otolaryngology-HNS, Ophthalmology, and Speech and Hearing Sciences. He is a faculty research affiliate of the National Primate Research Center, the Virginia Merrill Bloedel Hearing Research Center, the Center on Human Development and Disability, and the Autism Center at the University of Washington. He is also a faculty affiliate of the Center for Navigation and Communication Sciences at the University of Rochester and the Center for Integrative Brain Research at Seattle Children's Research Institute. He is on the Medical and Scientific Advisory Board of the Vestibular Disorders Association.

## **Kathleen C.Y. Sie, MD**



Kathleen C.Y. Sie, MD, is director of the Childhood Communication Center at Seattle Children's Hospital and professor in the Department of Otolaryngology at the University of Washington School of Medicine. A pediatric otolaryngologist, Kathleen Sie focuses on treating children with communication challenges such as speech delays and hearing loss. She started several programs at Seattle Children's Hospital including the Velopharyngeal Insufficiency Clinic, the Microtia Program and the Cochlear Implant Program. In 1999 when the hospital administration was planning for the new millennium, she proposed starting a Childhood Communication Center that would consolidate the efforts to help children communicate effectively.

## **Jennifer Phillips, PhD**



Jen Phillips earned her PhD in molecular genetics in 2003 and subsequently completed a postdoctoral fellowship in the laboratory of Monte Westerfield at the University of Oregon, developing zebrafish models of Usher syndrome. She continues to work with zebrafish Usher models as a member of the Westerfield lab research team.

## **Karmen Trzupsek, MS, CGC**



Karmen Trzupsek currently leads the Ocular Genetic Counseling team at InformedDNA. Karmen has been working in the field of ophthalmic genetics since 1997. Working first in a genetics lab at the Casey Eye Institute in Portland, Oregon, she helped work toward the identification of genes associated with susceptibility to age-related macular degeneration. After earning her Genetic Counseling degree from Northwestern University, Karmen returned to the Casey Eye Institute, specializing in inherited retinal and macular diseases and retinoblastoma. At InformedDNA, Karmen developed the first-ever comprehensive telephone-based genetics service for patients with inherited eye diseases. Karmen has served on multiple committees and advisory boards to develop guidelines and strategies for the incorporation of genetic testing into routine ophthalmology practice. She has a longstanding interest in Usher syndrome, and serves on the Board of Directors of both the Hear See Hope Foundation and the Usher Syndrome Coalition.

## **Jennifer Chao, MD, PhD**



Dr. Chao was born and raised in Santa Monica, California. She graduated with distinction from Stanford University and was awarded the Janet M. Glasgow Memorial Achievement Citation. She earned her MD and PhD at the Yale University School of Medicine, earning her PhD in Neuroscience

with a prominent neuroscientist, Eric Nestler, MD, PhD. Upon completion of her internship at Stanford, she continued her residency and vitreoretinal fellowship at the Doheny Eye Institute/USC. During her residency and fellowship, she was awarded the Fight for Sight Postdoctoral Fellowship, the Knights Templar Eye Foundation Research Grant, and the Heed Foundation Fellowship. During that time, she also initiated a research fellowship at the California Institute of Technology (Caltech) in the laboratory of Marianne Bronner PhD, a recognized world expert in developmental/stem cell biology. Dr. Chao has been on the faculty at UW since 2009.

Dr. Chao's research group studies inherited retinal degenerations and examines potential treatment modalities. Specific projects include the generation of stem cells from persons with inherited retinal degenerations (called induced pluripotent stem cells or iPSCs), generating retinal cells from these iPSCs, and studying the "diseased" retinal cells in culture. Ultimately, the group is focused on discovering new drug therapeutics that could benefit those affected by inherited retinal degenerations.



### **Mark Dunning**

#### **Chairman at Usher Syndrome Coalition**

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

## MEET THE FAMILY PANEL



### **David Hebert**

David Hebert is a professional firefighter/operator/EMT for the St. George Fire Protection District in Baton Rouge, Louisiana. He was born June 29, 1979 in Baton Rouge and lived his whole life in the small city of Plaquemine, Louisiana. While volunteering in November 2000, David discovered his love for the fire service. He went on to graduate with honors from Louisiana State University at Eunice in December 2011 with an associate's degree in fire science. After six months he was hired full-time by the Plaquemine Fire Department and ten years later he began to work for the city department of St. George Fire Protection District, as he wanted to take his care and influence to a higher level.

David met his wife Nichole in 1999. Married in April 2003, they are the parents of three children: Katheryne, born in 2004, Kesley, born in 2005, and Kayde, born in 2006. In 2004, Katheryne was diagnosed at birth with profound to severe hearing loss. After much consideration, David and Nichole finally agreed that Katheryne would get Cochlear implants. She was scheduled to be implanted a few months after her first birthday in New Orleans, but Hurricane Katrina soon put that on hold. Katheryne got her first implant in April of 2006, and after a series of setbacks with insurance she received her second implant in 2009. Katheryne was then diagnosed with Usher syndrome in 2011, and after getting genetic testing at Tulane University, she and her family learned she had Type 1b.





## **Ramona Rice**

Ramona has been living in Utah for many years without realizing that she has Usher syndrome Type II until the age of 42. She grew up as a hard of hearing child with no support or desire from her family to learn sign language with her until she had to take years of speech therapy to orally communicate with them. Ramona is a proud Usher syndrome advocate trying to make a difference for other Usher syndrome recipients, to know that they are not alone. She is a firm believer in meeting the challenge for fair and equal communication access in every single business available for our deafblind community. She created “Collaborative Partnership in Utah for People with Vision and/or Hearing Loss” strictly to gather as many community partners in Utah to support our blind and deafblind communities and deaf culture.

Currently, Ramona is a board member of Disability Law Center, and has been invited to join Utah Transportation Authority CAT committee team to help them to learn more about our deafblind community. She is also involved with Deaf Blind Advocacy Committee in Utah to bring in many blind and deaf organizations representatives to be educated on how they, too, can help their members with proper hearing devices, ASL/tactile interpreters, technology/print options, etc. One of her successful outcomes will be “Walk in My Shoes” book will be published this summer about our writers’ and their families who are affected by Usher Syndrome. She has been very fortunate to have a fantastic executive committee: Charlotte DeWitt, Karen Duke, Randy DeWitt, Rose Sarkany and Marisa Postlewater to make this happen.



## **John A. Romish**

John was born June 16, 1963 in Boston, Massachusetts. After John's first birthday, Children's Hospital concluded that he was profoundly deaf. At age three, he joined a nursery for hearing impaired children, where he learned to read lips. John attended the Boston School for the Deaf, the Beverly School for the Deaf, and was considering a signing-exclusive school when his family moved to Portland, Oregon. At sixteen, after John's teacher voiced concern about his vision, John was diagnosed with Usher syndrome. Counseling helped him and his family as they tried to understand and accept Usher syndrome and the challenges of being deaf-blind. John attended Cleveland High School and Portland Community College, studying in the field of printing. After graduating and working for a year, he went on a two year mission trip for the Mormon Church, which expanded his signing skills, added depth to his study of his religion, and gave him an opportunity to travel the country.

After leaving a manufacturing job, John sought out opportunities with the Lighthouse for the Blind in Seattle and joined, working in the Boeing production department. Today, he has been with the Lighthouse for the Blind for nineteen years and works using a computerized production machine. John has connections with his family, companions with whom he works, and his church community. The Lighthouse is a wonderful place to work for the deaf-blind, the deaf, and the blind because they provide opportunities for their employees outside of work. John is active in the Deaf-Blind community and around Seattle, which offers great tolerance and many opportunities to this community.



### **Moira M. Shea (Moderator)**

Moira 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 a former member of its board of directors. Moira also serves as Vice Chair on the board of directors for the Usher Syndrome Coalition.



### **Clare Weigel**

In 2013, Clare was officially diagnosed with Usher syndrome type 1B. Clare is from Tampa, Florida and is a recent high school graduate. She was a cheerleader for seven years and was captain her senior year on the varsity squad. In her spare time you can find her outside somewhere with her camera snapping photos, working out at the gym, or curled up in a chair with a good book and a cup of tea. Clare's plans for the future include her attendance at Samford University in the fall as an English major.

## RESEARCH PRESENTATION ABSTRACTS

### **A combined cochlear and vestibular prosthesis for the treatment of Usher syndrome: a clinical trial**

***James Philips, PhD***

Usher syndrome is often associated with both profound bilateral hearing loss and bilateral vestibular areflexia. The loss of hearing and balance function can be present from birth, as in Usher Type 1, or can be progressive as in Usher Type 3. When the hearing loss is sufficiently profound, cochlear implants can be used to restore hearing function. Currently, there is no approved therapy to restore balance function in patients with equally profound loss of vestibular function. Our group has been working on the development of a vestibular prosthesis, similar to a cochlear implant, designed to restore balance function in such patients. We have shown that the vestibular implant is effective in delivering vestibular information to animals and human subjects over several years. However, our original experimental prosthesis was not optimal for implantation in Usher syndrome patients because it was placed at the same location as a cochlear implant, just behind the ear. Therefore, if this technology were proven to be effective, Usher syndrome patients would be forced to choose between restoration of hearing function, or restoration of vestibular function. To address this issue, we have modified our device to combine both a cochlear implant and a vestibular implant in the same device. We have begun animal studies on the new device, and we are applying for modification of our existing FDA approval to expand our initial clinical trial to use the new device and include new patients with both hearing and vestibular loss in this first in man trial.

## **Hearing and Usher Syndrome: Diagnosis and Management**

***Kathleen Sie, MD***

In the era of universal newborn hearing screening, we are able to identify babies who are born with hearing loss (HL). Efforts to identify progressive childhood HL continue. With earlier identification of HL we have focused efforts on understanding what causes the HL for any particular child. The evaluation for causes of childhood hearing loss is rapidly changing. Hearing loss is frequently the first sign of Usher Syndrome. There are three main types of Usher Syndrome characterized by degree of hearing loss, vestibular and visual involvement. Clinical assessment and management of the hearing and balance symptoms of Usher Syndrome will be discussed.

## **Deconstructing disease genes: How zebrafish models of USH can help develop therapies for vision loss**

***Jennifer Philips, PhD***

Zebrafish have long been used to study vertebrate embryonic development and genetics. Recent advancements in gene sequencing and other molecular biology technologies now enable zebrafish researchers to create precise genetic models of human diseases. These models provide insights into what goes wrong when disease genes are inherited. The Westerfield laboratory has been studying Usher syndrome in zebrafish for more than a decade, and our efforts to understand the molecular underpinnings of this disease are now directed towards developing therapies to target the retinal degeneration experienced by people with Usher syndrome. Studying zebrafish that have the many different forms of Usher syndrome allows us to evaluate the similarities and differences

among the subtypes, and to test which types of USH might be improved by a given treatment. The ability to generate precise genetic models of pathogenic human mutations in Usher genes also allows us to test directly which therapeutic approaches will work on a particular population of Usher patients with the same mutation.

Some examples of our work include developing zebrafish models of Usher type 1F, which have a disease-causing mutation similar to that of many members of the Usher 1F community. We use these zebrafish to evaluate what goes wrong in retinas that lack normal Usher 1F function, and to test gene therapies for Usher 1F. Our zebrafish models of Usher 2A and Usher 2C, two genes that present challenges to developing therapies due to their large size, are informing us about which parts of the genes are most essential for normal function, knowledge that can lead to more targeted therapies in the future.

## **Unraveling Usher syndrome: The role of genetic testing** *Karmen Trzupsek, MS, CGC*

Genetic testing is becoming increasingly important for both patients affected with Usher syndrome and their health care providers. The most sensitive genetic tests can now detect the underlying cause of disease in more than 75% of patients. Still, most patients have not undergone genetic testing. This talk will explore the benefits and complexities of genetic testing, and provide practical guidance for patients and families interested in pursuing a genetic diagnosis.

# **Advances in Stem Cell Research for Treating Retinal Diseases**

***Jennifer Chao, MD, PhD***

The use of stem cells to better understand and treat retinal degenerative diseases, such as Usher syndrome, has been extensively studied in recent years. Both human embryonic stem cells and induced pluripotent stem cells (generated from patients) can be used to generate retinal cells, including photoreceptors (light sensing cells) and retinal pigment epithelial cells or RPE (support cells). These cells can be studied to better understand disease mechanisms and generate targeted therapies. Separately, stem cell derived photoreceptors and RPE cells can be transplanted in cell replacement strategies for patients who have lost vision. There are several ongoing clinical trials involving stem cell transplantation for patients with AMD, Stargardt Disease, and Retinitis Pigmentosa.

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