Transcript from the 11th annual USH Connections Conference, July 13, 2019

USHER SYNDROME COALITION

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Table of Contents:

WELCOME ADDRESS

Mark Dunning >> Hi, everybody. If we can get everybody to grab their seats, we can get started. It's going to take us a few minutes to get all of the interpreters set up, up here.

>> Good morning, everyone. We're just giving the interpreters a few minutes to get arranged, so we will be getting started momentarily. Thank you. All right. I think we're good to go.

Krista Vasi: Welcome, all of you, to our 11th Annual USH Connections Conference. I'm Krista Vasi, executive director of the Usher Syndrome Coalition. I look forward to meeting a lot of you that I haven't met yet. In this room you're surrounded by nearly 400 people who know what it's like living with Usher syndrome. If before this day you ever felt alone, know that you are now part of this incredible community.

A majority of you are here for the first time. Can I get a show of hands for the first timers? Wow. Oh, my goodness.

[APPLAUSE].

I really hope that this day will be as meaningful to you as it has to past attendees and their families. Before we begin, I would like to give some thanks. A lot of thanks, actually. First of all, thanks to all of you for coming, traveling from near and far. You have traveled from 33 states and three other countries to be here and we are so glad that you did. Thank you to our sponsors: ProQR Therapeutics, our silver sponsor. The McKittrick family, who has made it possible to provide scholarships to families to attend.

And our bronze sponsors: Akouos, Eloxx Pharmaceuticals, and Ush One See. I would like to thank our exhibitors and invite everyone to check them out during the breaks and lunch. Thank you to our volunteers especially Rachel Rabenn, studying genetic counseling with a focus on Usher syndrome. She is here to learn from you all today.

A big thank you to our speakers. We have a really wonderful program for you all. Thank you to our USH partner, the Usher Syndrome Society, for letting us host them today as they expand their global portrait campaign. You have seen the portraits lining the halls outside. They are powerful. You can be a part of that. If you haven't signed up to get your photograph taken yet, go see them in room 502

Scroll to top

during breaks and lunch. While you're there, be sure to say hi to Nancy O'Donnell, our Director of Outreach. She is right over here.

[APPLAUSE].

Nancy will be helping people join the USH Trust registry. You'll hear more about it later. Another thank you to our USH Partner, Ava's Voice, who is hosting over 30 kids and teens. Thank you to the Philadelphia Nanny Network for taking care of our little ones. Thank you to Clarke Hearing and Speech and Phonak for providing assistive listening devices.

One word about those. We had technical difficulties with that. If anybody here has a Roger pen that we can borrow, everyone can link an assistive listening device to the Roger pen. You can see the back room for it.

Wonderful, we have one person right here. We have people in the back that can help with that. And we can get everyone hooked up so everyone is capturing all of the information today. And a big thank you in advance to our hard-working interpreters and SSPs. A quick reminder for all of our speakers and everyone in the room, interpreters have breaks, so we might have pauses here and there as they switch.

Thanks to the Marriott for once again being a wonderful venue for this event.

Last but certainly not least, she will never admit this, but this day would not have been possible without Julia Dunning. Big thank you to Julia. [APPLAUSE].

A couple of other housekeeping items. The bathrooms are located, if you go out the middle room, middle doors, go straight out and then to your right. You will find the bathrooms. The guide dogs here, their bathrooms -- the guide dog relief room is actually in the corner, right across from the Salon D doors. And that should be all set for you.

I now would like to introduce the Usher Syndrome Coalition chairman, Mark Dunning.

Over ten years ago when Mark's daughter was diagnosed with Usher syndrome, he realized the importance of connecting with the Usher community. One of the most powerful ways we connect with those with Usher syndrome, with research, information and each other, is through these conferences. So please join me in kicking off our 11th annual USH Connections Conference and welcoming Mark Dunning. [APPLAUSE].

Scroll to top

>> Mark Dunning: Hold on. I got to get myself hooked up here. If I talk like this, can you hear me? Or do I have to bend down? Bend down, okay. All right. Great. This is sort of the way I live my life, bending down like this.

Thanks, everybody. Thank you, Krista, for that nice introduction. My name is Mark Dunning. I'm the chairman of the Usher Syndrome Coalition and one of the founders of the Usher Syndrome Coalition. I'm the father of a soon to be 21-year-old daughter who has Usher syndrome.

And I am also the father of a soon to be 18-year-old son who also has Usher syndrome -- sorry, you do not have Usher syndrome. Do you have Usher syndrome, Jack? No. He is helping out with the kids today. You'll be able to recognize him because he is almost as tall as me.

He claims to be taller than me. We know that would be impossible because there is no way my little boy is taller than me. Bella is not here however, which is good. Because it gives me an opportunity to talk about her without her stopping me.

And you know I want to tell you about Bella for two reasons. First is, she is my daughter and I have a room full of 400 people and an opportunity to brag about her. Why wouldn't I?

And then the second reason, is because we have almost 50 kids here today and we have more than 50 sets of parents here today and you saw for the first time, the hands that went up earlier.

And we have young adults here today, who are dealing with all sorts of different challenges, including social isolation and we have a number of older adults here today, who have never met anybody else with Usher syndrome.

And the truth is there is -- just about everybody here today is afraid, in some way or another. They're afraid about what the future might hold for themselves, or for a family member. And they're afraid that they maybe cannot or will not ever be happy. And believe me, I understand. I started this organization because I was in a panic about Bella's diagnosis. And you know, I am still terrified about her losing her vision.

We're all here looking for hope today and I think you'll find it. In fact, I know you will find a lot of hope here today. You probably already found some hope just by connecting with some people that you have met here today. You're going to hear some inspiring stories with people with Usher syndrome, including our yellow dressed guy up here who I am sure you have all seen so far.

Scroll to top

And you're going to hear about science and the treatments that are under development.

And you will not be alone today. Today people with Usher syndrome are the majority. And that is a rarity for people with Usher syndrome. So enjoy that today.

We built this Usher syndrome community, and when I say we, I mean we. The big we. All of you guys. And we have all built this Usher syndrome community to provide hope to families. And one of the greatest sources of that hope is to find treatments. We need your help. Krista mentioned this before. The goal of the Usher Syndrome Coalition is to identify everyone in the world who has Usher syndrome and get them into our registry USH Trust and get them genetically tested.

You can do so today. Nancy is over here. She will do jumping jacks so you can see where she is. She is a grandmother. That's impressive. She is fluent in English and ASL and she will be over there. Please grab her. And get yourself registered if you are not already registered. We need you to do this urgently. We are regularly contacted by researchers who are researching Usher syndrome.

They contact us for two things. One, they need clinical trial candidates and particularly for the pharmaceutical companies, they need a market to deliver the treatments in which they develop. And you guys are both of those things. I have one hand. So I will throw it over my shoulder.

So now, look. If you register it does not mean that you are volunteering for a clinical trial. It just means that you will be informed of clinical trials that are occurring. Okay. And I can tell you beyond a doubt, that we have seen more activity with pharmaceutical companies in the last year, than we have seen in the last ten years combined. There is money out there. [APPLAUSE].

It's excellent.

There is money out there that is looking to invest in treatments for diseases. And we are on the short list of the diseases they are looking to treat. And the best way for us to encourage them to keep us on that list and move us up on that list is having a robust registry, and be an easy contact for people with Usher syndrome. You guys are the key to that.

If you are interested in helping to support our efforts, we can always use both volunteers, and also financial support to continue this stuff.

Scroll to top

One of the best ways to do that is to sign up to give monthly donations to the Usher Syndrome Coalition. It just comes straight out of your credit card or bank account. So if you skip a pizza for a month or want to skip a night out for a month, that one goes a long way towards helping us to find people with Usher syndrome.

Now, I told you that I was going to brag about my daughter but first I want to introduce our next speaker who is Dr. Marly Kenna, a graduate of the University of Philadelphia. She has Pennsylvania connections. I want to make sure I get this right. Director of clinical research at the Boston's Children Hospital, otolaryngology and hearing loss there. It's impressive and well accomplished. I found it amazing that she found room in her life for me and my dopey dad and family, among the thousands of patients that she has treated. But she did.

And now, to me, as I know to many of you in the room, she is not Dr. Kenna, but Marly. It was actually Dr. Kenna who kicked off the two worst days of my life. Thank you very much. It was Dr. Kenna who told me that my then infant daughter was profoundly deaf.

And eight years later, it was Dr. Kenna who told me that the cause of my daughter's deafness was this disease called Usher syndrome.

And you know, it speaks to her character and her kindness that Marly became family after those interactions and I don't know how I would have survived those days without you.

[APPLAUSE].

Marly's commitment to families, the families she treats is just tireless. I mentioned I was one of the founders of the Usher syndrome. Marly was one of the other founders of the Usher Syndrome Coalition and everything that happened here in the last ten years or so as we have been running the Coalition, has Marly's fingerprints all over it. Growing the research collaboration with a few researchers talking to each other, and at the last symposium, we had 125 researchers there.

And last year, you know, and our outreach now reaches people around the globe. I was just introducing myself to a family from Israel.

Krista mentioned we have a number of different countries represented here. Last year Marly helped to organize the symposium that we ran in Germany and just in the last week, we have been contacted by deaf-blind organizations in India and Nigeria that want to collaborate with us. We truly have a global outreach and

Scroll to top

global reach here both within the Usher syndrome community and the researchers. A lot of that has to do with Marly's work. If you're in this room, I promise you she had a positive impact on your life, whether you know it or not.

I know she has had a positive impact on Bella's life. This is where I get to brag about Bella. She is not here today because she is working at a dude ranch in Colorado for the summer. She is living independently in the middle of nowhere. And she helps with the shopping, cooks the meals, helps to maintain the ranch, helps care for the animals, does chores and she and her friends go out on the nearest town which is 45 minutes away from where she is staying. And she doesn't call nearly enough because she is having way too much fun.

Next fall, Bella will start her junior year in college by studying "abroad" in Hawaii. She is majoring in biology, has very good grades in college and has a very good social life.

Most importantly, Bella is happy. Really happy to the point that she never calls anymore. And Marly had a lot to do with that. Because she gave our family hope when we needed it and Bella has built her life on that hope, that belief that she can have a happy future and I know when you hear Marly speak today, you will feel that same sense of hope. That's really what I wanted to tell you about Bella.

I wanted to tell you about a young woman with Usher syndrome who has profound hearing loss, yet is majoring in biology in college and is spending her days taking questions on the floor in Colorado from guests at a dude ranch.

I want to tell you about a -- interpreter switch. Excellent.

I wanted to tell you about a young woman with Usher syndrome who has poor balance and yet spends her days riding horses through the Colorado Rockies. I wanted to tell you about a young woman with Usher syndrome with vision problems, yet guiding tourists through rough terrain on horseback. Yes, that might sound scary. She is good at it. And importantly, I want to tell you about a young woman with Usher syndrome who is happy.

I want to give those of you who are struggling with the belief and understanding that it's impossible, it is. I want to give you the hope that Marly has given to me. Happiness is available to people out there with Usher syndrome. And you will hear that a lot today. And you will hear a lot about the hope today.

So a few years ago, we established the Foresight Award for recognizing people with untiring dedication and commitment to the Usher syndrome community. Academy awards have the Oscars. We should probably call this thing the Marly

Scroll to top

because she embodies everything that the award stands for. It's my honor to present this year's Foresight Award to the next speaker and my friend, Marly Kenna. [APPLAUSE].

Smile because somebody is taking a picture probably.

So we're going to switch the Rogers pens so we can move around a little bit easier up here. If you want to give us a second to get that going and then Marly will speak.

"Treatments of the Future for Usher Syndrome: The Future is Now"
Margaret Kenna, MD, MPH, Keynote Speaker

>> Can you hear me? I'm not as tall as Mark. So I don't have to hold the microphone. So I am obviously Marly Kenna and I had had no idea about this award. But I feel really lucky to be here and I know many of you. I am hoping to meet many more of you. My life is definitely richer because of all of you. I'm going to advance the slides and look at the slides at the same time. Hopefully this will all work out. I have the honor also of being the Sarah Fuller Chair for Hearing Loss and Hearing Restoration at Harvard. When I received this honor and my boss named the chair, hearing loss and hearing restoration, I said you've got to be kidding me. There is no way we can do that. He said we can do that. We are giving you this title, you have to do it. Sarah Fuller was a speech and language pathologist and her family worked with us and endowed the fund. I'm proud to say we're making progress on the hearing loss restoration front, as much as the research informs the area of vision impairment as well.

I have nothing to disclose. These aren't advancing. I'm sorry. Oh, there we go. Also there are a lot of people who helped me with this particular presentation. I'm an otolaryngologist. And people that I have worked with provided slides, information, provided articles and gave me support to put this together. Gwen Geloec, Heidi Rehm. Andrea Oza. David Corey, researcher USH 1F. Eric Pierce, you know, and an ophthalmologist, Anne Fulton, who works with our patients and of course our families. So how I did get into this to begin with? I'm an ear, nose and throat doctor. As Mark mentioned, we have been able to identify children earlier and earlier with hearing loss because of hearing screenings. We now identify babies at birth who are deaf so we meet those babies in the ear, nose and throat department. Talk about managing their hearing loss, and we meet them young to figure out the reason they cannot hear very well. One of the more common causes is Usher syndrome. And now we

Scroll to top

have genetic testing for this and we identify these babies in the ear, nose and throat department, rather than as adolescents or adults in ophthalmology.

I'm going to talk about DNA because it has to do with therapies down the road and speakers who come after me to talk about this really exciting stuff. Everybody knows that we have DNA. There are about 20 thousand genes. There are plants that have more than we do. Our genes are more important! We have 67 million miles of DNA in each person. A lot to work with. Most of the DNA for everybody in this room is identical to the DNA of everybody else in this room. Half a percent is what makes us different and has to do with how we hear, how we see. If you like coffee, all of these things are genetics.

23 chromosomes, the ones on the bottom right X and Y and everything else, chromosomes that carry genes for hearing loss, vision impairment and as I mentioned, coffee consumption. And the thing that is cool about DNA, all of this is squished into little tiny packages and it gets copied to make new humans or new proteins, the DNA stretches out, in each cell stretches out 6 or 7 feet. That's one cell. All of the DNA in our body, if we allowed it to go to the moon and back, 300 thousand times so if we can just harness that, we would not need falcon heavy rockets. So this is how DNA is stored. On the top left is one of those little puppies that they used to make toys when I was a kid. A slinky.

Everything is squished together. The cool thing about the dog, you can see the important part of the slinky is stretched out. Turns out that DNA is exactly the same thing. And in thinking about how this works, the top sort of spiral is DNA.

There are a lot of copies and they are spiralled together and that was figured out in the early 1950s. Not that long ago. Kind of when I was a child. That gets copied and becomes RNA. There are a lot of different types of RNA and the RNA gets copied and turns into proteins. When you're thinking about how you're going to alter the molecular make up of the human, you can alter the DNA.

You can alter the RNA or you can alter the proteins. If you alter the DNA, it's theoretically permanent. And if you alter RNA, it gets made all of the time. A lot of therapeutics that are discussed today, look at both DNA and RNA. Either way, if you change either one of those. You can change the protein. What you want to do is make a protein that works better.

That's really in the long run what we want to do. However we do it, we want to make a protein that works better and there are a lot of different ways to do that. There are a lot of different ways that DNA is damaged, malfunctioned or just does not turn out right to begin with.

Scroll to top

There are different types of mutations and for each type of mutation, there is a different strategy of how to fix it. It makes it complicated to figure out where to attack. This is a current list of the Usher genes and where it was identified. And this list is -- sorry. This list is in constant motion. On the left-hand side, the locus where the gene is located and the right of that is the actual gene and then to the right of that is the year it was discovered. So the first Usher gene, Usher 2A was discovered -- I'm sorry, first Usher gene discovered 1B. MYO7A. 1995. Really not that long ago. There were also genes thought to be Usher genes but they aren't. This really shows the complexity of trying to figure things out.

And so when we say somebody has Usher syndrome, what we mean is that they have a hearing loss, and have or will have vision impairment. There are lots of other things that cause hearing loss and vision impairment together. One of the important things about genetic testing is making sure that we have the right gene. If we have the right gene, we can come up with the right therapy.

The last thing we want to do is treat for the purple gene and they have the red toe nail gene. It's really important to figure this out. So one of the common things about otolaryngology and ophthalmology, genes are the same for Usher syndrome and work in various different locations. Looking at the slide, to the left is the photoreceptor, the genes -- the cells in the eye that actually convert information to light and right is the hair cell in the inner ear that converts information to sound so different Usher genes are situated in different parts of the cells and they all work together like dominoes that go forward. If you take one in the middle out, the rest of the dominoes don't make it to the end.

The other thing that is tricky about photoreceptors in the eye and hair cells in the ear is they are what's called terminally differentiated which means when they get to their mature form, they don't reproduce again. It's not like a liver where you damage the liver and liver makes new cells or like skin, you get a sunburn, and it falls off and you get new skin. You don't get new photoreceptors or hair cells. Anything that we do to change what's going on in the cells could be permanent or long lasting.

It changes the way the research is happening in this field as opposed to other fields where the cells are turning over. Current therapy for Usher syndrome is Vitamin A, wearing sunglasses, antioxidants, fish, salmon, cochlear implants. We know people are wearing them. Retinal implants. Vitamin A is good up to a point and sunglasses will change it up to a point and we know that antioxidants may slow things down. They don't treat the disease. They help and they're good but they don't treat the disease. So Bill Kimberling, one of the founders of the Usher Syndrome Coalition and part of the team that identified Usher 1B, MYO7A,

Scroll to top

Since that time, they're able to correlate genotype with phenotype. Find or develop animal models. Elucidate disease mechanism. Screen the human population to identify people like the USH Trust and test the treatment in these folks.

If it works out and it's safe, you can roll it out to a larger population and of course they test the treatment in animal models.

If you look at clinicaltrials.gov, not just gene trials, there are 17 studies under Usher syndrome. However, most are completed, not recruiting or they have terminated. The ones that are not terminated are ProQR, USH2A. Scotts 2, stem cell ophthalmology study. No information about that. And all I would say is I would be cautious before entering this particular study and there is USH Stats that are not recruiting new patients.. That's it. There are a couple of studies. A few years ago when we were in Germany, there were no studies except for follow-up for the USH Stats. Things are improving. What are the strategies for gene therapy? Correct the gene. Replace the gene. Move it around. Restore function that isn't there. Override abnormal or bad function or inhibit abnormal function. Those are all different strategies depending on the gene you're trying to fix. And there are different techniques for gene therapy. Everybody has heard about CRISPR. It's basically cutting out a piece of DNA and slicing and putting a new piece together and hoping it works better. It's a new piece of technology. Exciting. But it's not foolproof. Replacement genes that you can actually package inside viruses. You can skip exons. If exon is part of the gene, you can skip over the exon that is the problem and just go on. And then there are some other things that people are working on.

Stem cells, nanoparticles and small molecules. Basically what you're trying to do is trying to make enough of a protein to work. Does not have to be all of the protein. Just has to go from protein that is not working, to some protein that works. Some might be good enough. There are viruses that are utilized and put the piece of DNA or RNA that we want into the virus, like a ship going off into the moon into the patient and there are several different viruses used. Adeno, retroviruses, and it has to get to the patient and attach to the cells that you are interested in. If you want to send a gene to the cell that make the vision or hearing better, you don't want them to go to the liver. We don't care about the liver. We don't want it to hear or see. We want the eyes and the ears to hear and see. You have to figure out where it's going. There has been some exciting work. USH 1C makes a protein called harmonin, able to take an antisense nucleotide and it affected RNA to produce protein production. Using that same model, able to have the adeno virus. That was used in our laboratory. There is

Scroll to top

also USH 1G. The protein is called sans where there is partial restoration of hearing and balance published last year and whirlin USH21D, able to improve the hearing. There are a lot of things on the hearing side that hopefully is applicable to the balance side. Sorry. This is delayed. All right. Sorry. Just enough in the way if you push the button. All right. Anyway, suffice to say, this was Jen Lentz's work. Here it comes again. Took mice and created model of USH 1C to population in southern Louisiana. A single mutation of the patients and was able to restore hearing and balance in these mice very reproducibly. Now working on the same model looking at the retinal phenotype in the group of patients. Recent USH 3 work, looking at chemical stabilizers, USH3 gene and a corollary group identified Clarin-1. The ribbon synapses between the hair cell and the brain. Turns out it may be another focus of work to allow people to hear. And in the eye. There may be a great way to translate the ear to the eye. Gene therapy approaches. The eye is very susceptible. What you do into the eye does not necessarily leak out into the rest of the body. It's very important. You can monitor it non-invasively. Check vision for example without sticking needles into peoples' eyes. The very first, USH stat was a lengthy virus delivery of MYO7A, USH 1B, injected into mice, and phase two, people. Being followed up now. Everybody in the room now in 2017, Luxturna, basically received FDA approval for the treatment of RPE65 gene abnormalities. It is not Usher syndrome but this is an excellent model for gene therapy. Took them 20 years and there was an actual dog model that allowed them to study this beyond mice. This is the eye obviously. Two things you can inject things that affect vision. One in the retina, way in the back of the eye and the vitreous, the fluid in the middle of the eye.

Usually this is done under sedation or when the patient is asleep. The eye is right there. As long as it's done by people with good skills in the eye, relatively straightforward. Sorry. A little delay. This is a slide from Spark Therapeutics, the makers of Luxturna and the picture on the left, Jean Bennett, University of Pennsylvania. Extremely exciting work. Hopefully we will be able to build on this for Usher syndrome. This is the dog model.

Naturally occurring model of the RPE65 gene. Going forward in humans. The very first human trial of this was announced in the New England Journal of Medicine in 2008. It was three patients.. They were very cautious about this. And in 2017, FDA approved this. You can see there was some lag time to make sure they got it right and now both adults and children have access to this.

One of the down sides is one injection for one eye is 450 thousand dollars and obviously as time goes on, that has to improve. The dog got it for free. I think he

Scroll to top

got it for free. There are a lot of therapeutics. Pro-QR. RNA based therapy. Working on LCA10, Leber. And talking about therapy For Usher 2A. I'm not going to talk about it. They will talk about it. There are other people interested in this space. Mark said people are calling all of the time looking for new therapy. Editas, Eloxx, you will hear from them. And there are several other companies working on this. Very exciting and I couldn't have made this slide last year. The companies were not ready to talk about it. I will skip ProQR because they will talk about it and there are several studies. Mass, University of Michigan, Oregon, Casey Eye, and several other locations and they are on the ProQR website. This is technology. Exon skipping. Basically you take a piece of the DNA that contains the mutation and you basically skip right past it and use the rest of the DNA, RNA to make a protein that allows you to have a better working protein. Sounds really simple. And it's not so simple.

But not impossible. But not simple. Eloxx is using a different strategy where they are trying to go around the premature stop codon. All DNA has pieces that say when to start making RNA or when to make a protein and when to stop. Sometimes the stop is in the wrong location. It's like that thing of dominoes. One domino in the middle prevents you from going forward and their strategy is to try a read-through. Basically pass that stop, go to the correct stop and make a protein.

So there are some challenges to gene therapy. There are multiple types of mutations. There are multiple genes. Usher genes are very large so whole genes do not fit into the viral packages.

Each gene often results in several different proteins and they may be at work at different parts of the photoreceptor or hair cell. You fix one and may not fix the other but you actually need both proteins to make things run effectively. I did reach out to a lot of researchers doing this type of work and they provided me very vague updates. Eric Pierce, a pediatric ophthalmologist, many of you know him. Working on an addition to the ProQR, additional Usher 2A projects. David Corey and Harvard medical school is using the zebrafish because mice are too big and we're going to zebrafish. A great model of the eye. Jen Philips and Monte Westerfield developed this. I think it's going to be exciting.

Basically in summary, and I am very sure that I have missed things and I hope I did not mischaracterize any research, USH 1B, USHSTAT are being analyzed and 2A you will hear from ProQR and Eloxx. ProQR is recruiting. And for USH 3, they are working on molecules in the laboratory. 1F, we talked about zebrafish and for USH 1C there is a mouse model and soon to be a pig model in development of USH1C. As Mark said, there is no reason to do research if we

Scroll to top

can't offer it to patients. So join the USH Trust. Thank you very much. [APPLAUSE].

I will be around. If anybody has questions about any of this or if I got it wrong, please let me know. Thank you.

>> We're going to give a two minute break between speakers as well.

>> Okay. Where are my interpreters? Thumbs up? Are you good to go? Okay. We are good to go. Next speaker is the guy in yellow right here Dario Sorgato. He is living in Berlin right now, but researching in Italy. I met him last year. He has been a great advocate for people with Usher syndrome around the world. We are glad to have him. He has all sorts of stuff he is trying to figure out up here some technical difficulties.

>> Let's give a nice big round of applause. Thank you. [APPLAUSE]

“Usher Inspiration: From Dreams to Action” Dario Sorgato

>>Dario Sorgato: You warm me up. Yeah, Mark already gave an introduction. Here I am. I am Italian and 41 years old and live in Berlin as he said and I was diagnosed with Usher syndrome when I was 17. For those who can see me, I am completely yellow. You might have seen around, this guy going yellow. I did it because in Italian, it's giallo and it rhymes with beautiful. Yellow is beautiful. And you won't miss me for the whole day. If you miss me, I will find you anyway.

That's why I will tell you the story of why it's yellow but this is the fun part. Yeah. A little more about me. I'm a designer. Study design. A blogger, vlogger and social activist. The main thing about me is I have a great passion for traveling.

I still have to learn this thing. Travel is nothing but the most peripheral shock wave of a deeper motion, like the outer circle caused by a stone thrown into the pond.

I love this sentence. I believe that every great traveller has internal movement to look for something, look for more and for me it's the same. I'm an inner motion and I don't know exactly where it's coming from. It's most likely related to Usher syndrome which is causing me this curiosity for the world and the need for searching, for looking, for finding and meeting new people, new countries and new places.

Scroll to top

I believe that every great traveller must have this inner motion. I like this image. Because I believe traveling is just the outer circle. I could manifest this in another way which is writing a poem, a painting or so on. But for me, it is traveling and that's why I go travel the world and this is a couple of photos of myself in front of the Kremlin in Moscow and the Great Wall of China and I am putting my hand by my eye and behind the ear. It's representing Usher syndrome. And I go around the world, taking photos of me and sharing them around to see that we do it for other disabilities. The pride of disabilities. Deaf pride and so on. Why not Usher syndrome pride? It's hard to say, but let's try. [APPLAUSE].

Thank you.

I will share some of the main trips and travels that I did. This is one of the first big ones between 2004 and 2005, I traveled to Australia and New Zealand for a year. Initially there for an internship and it turned out that I traveled most of the country and this van, of course not driven by me, I did 100 kilometers with this van. My friend was bitten by a snake and had to drive to the hospital.

Drove 100K an hour. I don't know how much that is in miles but it was fast. For a -- can I say shitty van like this? And I published a book about this trip called "A Year In Eight Hours" and I thought wow, this is a dream. If you have a good sleep -- not like last night -- but if you have a good sleep, eight hours is like a dream. In Australia, dream time is the time of creation. For me, it was my time of my creation.

That's a long story. I have a book about it. I don't have time to talk about all of it now and then there is Camino De Santiago. You may have heard about this by now. 800 meters that goes through Spain and I didn't have time to complete it in one trip. This was in 2006 and 2007. I published a book about this as well called "There is No Time" because I believe we need to be slow -- who am I talking to? Usher syndrome say this to people. But we are slow anyway. If we perceive walking, it's a way to perceive life, nature and the surrounding better. Because we can perceive the details in a better way. This is life and trying to go beyond life for our society. Another important trip was between 2008 and 2010. That's nearly two years I was on board of this vessel. It's a Chinese vessel with square sides, and a red deck. It's called the Greek philosopher. I sailed from Cape Town to Havana. Two months of crossing to De Janeiro. It was a trip that was hard to believe that I was on this boat. And of course it was life changing. Because this is the moment when I accepted my disability.

Finally I had to face the limits. And I had to tell others that I had vision impairment and hearing impairment, and on a boat, if you don't say it very clearly,

Scroll to top

you can put other people in danger. So I had to be very conscious about this. That is what I learned to accept my limits. Even in this case, published a book called "Acqua" meaning water. A journey. And this is mostly a photographic book. Indeed, this trip, I accepted my limits and founded a project, initially was just a blog, where I was posting articles about vision impairment, hearing impairment, perception and so on called NoisyVision. Today it's registered in Italy. We have the pay off, we don't see the problem. We need to laugh about our problems and difficulties because we have to take it with a smile. We have to take it with happiness. And yellow. Yellow is a color of happiness, passion, joy. Something nice and beautiful. So we have to show this part of us. Even though we are facing difficulties, we have to try this way.

In 2013 I organized award show with founding European Commission. 60 people from all over Europe came to Berlin and did a worship about accessibility. And we went to investigate the city, understand what is good and what is not good in terms of accessibility for people with visual impairment. Mostly visual impairment.

And out of this worship, we followed the yellow. Some of you might have seen this already. Stands for less pain to the world, yellow. So it's about joy, passion and so on. But also the part of accessibility. Because if the world would be more yellow, it would be more accessible. Think about the steps when you go downstairs. If they put the stripe on the first and last step, it would be easier for you.

Think about the sidewalk. It would be easier for you. Think about the gray background in the buildings. If they were yellow, we want hit them. So a world with more yellow would be a more accessible world, to talk about accessibility. At the end of the day, problem with the world is not that we have limits, it's just mostly that the out world is not ready to take us.

Accessibility is the main topic to make our life easier. That's why it is important to advocate and fight for more accessible world in terms of communication, information and in terms of navigation. This is a map of the world -- oh, I'm not going ahead actually with the slides. Sorry. I have only one brain. This is the map of the world. Yes, and there was a lot of yellow dots. Everywhere that people go, they maybe take a T-shirt or something to make a funny thing. This is a friend of mine. Went by Mount Kilimanjaro and all of the people, the map of the world, you have seen before, and we tried to make this map a little more yellow. And if you want to join this campaign, you are welcome to do the same and use the #yellowtheworld.

Scroll to top

Here we go. To make it really big. I decided to go to the highest point on earth, the yellow symbol. It means we like it yellow. Where else could it be that everybody else can see? On top of Mount Everest. In 2015 I said I want to go to the mountain. I went to the base camp. Not to the top of it, not yet. And these are photos of the children. The campaign. And I went to share with the people around. This campaign of yellow the world. And finally made it to base camp. This is a photo with Everest in the back. A little lower. 4000 kilometers above sea level. That's the photo I was talking about. I made it.

Made a little movie about this. You can find it on YouTube. Actually speaking English in the movie. Yellow the world, solo expedition. And after all of this, I said, I want to share this with other people.

In 2016, I organized a trekking. The Gods like it yellow, from Bologna to Florence. I should have measured this. But I took 12 people the first time, blind and visually impaired for this trekking. I wanted to share this adventure. We don't need to but it's great to lead life as an adventure. You want to do something that makes you feel happy and passionate. In 2017, I did another worship similar to the one in Berlin, the yellow Reykjavik. Went to the city of Reykjavik and the same year, went with a group of blind people to volcano Etna in Sicily. It's all yellow. Very yellow already. But anyway, it's a great experience.

This is a photo of me with a blind lady and there was a dog with asthma. Unfortunately in this environment, the landscape, not accessible for dogs. In 2018, did a ride, tandem and took 15 people around Berlin with Usher syndrome to enjoy the bike.

Sometimes we're not able to do this and it's nice to give a chance to people with visual impairment, a chance to ride a bike and this year I did trekking from Piazza. The donkeys need to be included as well. Fantastic trekking. It was only four days. With this trekking, we won the award of accessible touring, because it is something that is really fantastic, to give a chance to people with blindness to enjoy the nature. I would like to say the people who enjoyed it more are the non-blind people. We can teach them in a way -- let me use the word teach, to see the world without senses. They can learn from us to perceive nature in a different way that they have never experienced before.

Because people who see, they think with everything and enjoy everything just because they see. This is not the point. We have a unique point of view.

A unique way of perceiving the world which is our power. Here we go. We have the Connection Conference and next August, I will go on a sailing boat from Naples. A week-long journey. Mostly I have been talking about mountains. But

Scroll to top

we want to conquer with yellow, also the sea. You might say at this point, wow, that's so inspiring. Can I hear that all together? Wow! That's so inspiring!

Yeah. It might be. But you know, what about me? You say okay, this guy has been up and down the mountains, go to the sea, here and there. What about me? Like I was saying, the power of Usher syndrome is that we're aware of our unique perception of the world.

You don't need me to tell you that. We need to tell other people and need to share the uniqueness that we can have. That's how we can turn our weakness into something powerful and strong. We need to educate our mind to empower details of what we see and hear. Might see very little, and hear very little, different from others, but what we see must be -- it's at the end of the tunnels which we have in the middle, a flower, bird, sunset, maybe it's blurry, maybe it's dark.

Do we see with the eyes? Or do we see with the heart? That's what we need to educate. To transform what we see, into something for the soul so the senses are channels to provide information. And we can be an inspiration to other people.

This is the first time I'm with such a huge Usher syndrome audience. Usually when I share the story, the people are -- let me say the word normal. It means nothing normally but I want to say not visually or hearing impaired. They say wow, that's inspiring. And that's actually our audience when we do something. Because it's not only within ourselves. This might be the story and what I am sharing today, might be something that can trigger something today. Our lives are already unique and I don't want to deny that Usher syndrome is surely a burden in our life. But can it prevent us from dreaming? Are we dreaming with our eyes and our ears or are we dreaming with our mind and our heart? The dream does not have to be extraordinary. Our minds are exceptional just because we are unique. We need to really understand to make this our way of thinking.

And we are not unique because we have Usher syndrome. We are unique just because we're people. If we're the sun in a solar system, Usher syndrome is just one satellite. One day it might be Mars, Venus, Jupiter, Pluto. But we're just one satellite. We're not what we see in here. It's what we are. This is something that we really understand, and make it as our way of living.

So, now, how can we turn dreams into action? I don't have the solution, but Google has it.

Scroll to top

You go to Google, how can I turn dreams into action? Google has the answer. It's really true. You can Google, how can I turn dreams into action and you can get a list of things of how you can do that, if you need to do that.

There are a lot of people that do this as a profession. So I am not here to share this with you because Google is doing a better job. However, if you Google, how can I turn dreams into action if I have Usher syndrome, you get no answer. So Google is not God and doesn't have the answer for us. Neither do I.

But I want to share some tips which I got from my personal story. First of all, do you have a dream? Although they say it's better to have a clear dream, clear goals before you start, I don't agree. I think you have to start somewhere. Somehow. Dreams will take a different shape as you go along. Start somewhere. It might be a small dream. Might be big, but just go. The other big problem, money. I don't have money.

Don't hide behind the excuse, I don't have money. I know it's difficult. I don't have money myself. But I found ways and there are plenty of ways to find money. It's more difficult to find the dream than money. Trust me. I don't have time to share a lot of information on how to find money. But I'm happy to share, what about time? Set priorities for what really matters to you. We all have a job, different things to do. We take care of our children, parents and so on. But it's about priorities. Sometimes we need to do an extra effort to pursue our dreams. So even this, time and money, I'm sorry, but I don't accept excuse that I don't have time and money to realize my dream. Another important point is to connect with others.

We are a big community and need to go beyond the region. Because what you can learn from other countries and other ways of thinking, in this case, beyond the states or beyond the Western society and so on, you can get it from others. So it is important to stay connected. And therefore this conference and this room and this situation is so important.

A bit selfish but you should do things you like. A lot of people know this. And they say we fear the judgment of other people. But we have to do what we like. And we have to put the center on our important things. Research, study and learn. If you don't continue to research and to learn, you will not succeed. There are countless available tools to support your dreams. You have to dig them out. Internet. Other friends that you connect with. There are plenty of ways to keep learning, and we need to improve the information that we know about certain things, in order to achieve our goals.

Scroll to top

I also started a lot of time with my dreams from the local communities. For example, the base camp, I did a crowd funding campaign because I did not have money to go there. The first to help are friends and family, the community, they can trust you and can be supportive. I'm not talking only about economic support.

Sometimes we need emotional support, other motivation and so on. Don't wait. Is it a bakery? Coffee shop? Cycling trip? Parachute jump? Today is your new tomorrow. I hope you come out of this room and say today is your new tomorrow. Please reach out. I will share my contact information. And you can reach out to other organizations, support, and online communities. There are plenty of Facebook groups and other communities out there where you can find help.

The question comes, am I there yet? You heard the story. You have achieved the dream and so on. No. Lucky I'm not there yet. It's a journey. I want to continue this journey.

We are there today because we want to be in this journey together and travel together. Some dreams might be along with yours and we can share this journey together. I believe that traveling in a group is especially in the nature with other Usher people, it's a life changing community. As I said before, continued to do this trekking with the donkeys and Neptune and so on, but I believe the best way to perceive the world is to understand that we might have difficulties seeing and hearing but again, we shouldn't have difficulties in dreaming and dreaming big! Thank you.

[APPLAUSE].

Now one last thing. I want to share with you this slide -- oh, no it's gone already. Yellow the world is also a nice way of shouting all of the emotions we have. Now I'm going to do this with the biggest community ever.

When I say three, you will say yellow the world. It's loud. One. Two, three. Yellow the world. One, two, three.

Can be louder, okay. You can be a little louder. It's still morning. Still waking up. Let's be very loud. We want to show that we are proud and happy. One, two, three. Yellow the world!

[APPLAUSE].

>> Thank you, Dario. That was fantastic. How about another hand for Dario?

Scroll to top

[APPLAUSE].

We have to do just a couple of minutes of rest for the interpreters and some technology issues so we're just going to take care of that. >> so interpreters, just give me the thumbs up when you guys are ready.

Okay. We are just about to get started. So our next speaker, has the difficult job of following Dario up here but even though she probably won't be quite as dynamic of a speaker that Dario is, the stuff that she is talking about is really important to a lot of people in the room. This is Dr. Susan Schneider, of Eloxx Pharmaceuticals from U.S. based and Israel. Where are my Israel people? After this talk, connect with her. She knows retina specialists in Israel that she could put you in touch with as well. So Dr. Susan Schneider. [APPLAUSE]

Scroll to top

“Establishing a translational read-through approach for inherited retinal disorders” Susan Schneider, MD

>> Susan Schneider: Thank you all very much. It's really a pleasure to be here and it is my first Usher meeting. I have taken away two things so far. One is happiness. I have seen so much happiness around me and the other is yellow. I'm going to show you a little bit about how to be more yellow at the end of my presentation. And I am already learning a lot from you as well. I have a friend in the audience teaching me how to sign. For him and for all of you, I am ready [verbalizes and signs “I am ready”]. With that, we will go ahead and get started and for our Israelis in the audience, Shalom.

So on behalf of Eloxx Pharmaceuticals today, I would like to speak with you about what we're doing in development space. We're not quite yet in the patient space in the clinic but with that, the other thing I would like to have you help us with today is with these patient registries.

Please get sequencing if you can and join the registry so when we're ready to get into patients and clinical trials, we know where you are and if you want to participate, it will make it a lot easier for us and for you to move forward with the clinical trials.

Because we're a publicly traded company, I am obliged to let you know these are forward looking statements.

So the reason I joined this company is because people are blind and there are no treatments. I don't have to tell this audience that. It's really a passion for me. It's a way that I can help people move forward and I am hoping that what we have to contribute will help all of you.

When we talk about large unmet medical needs in inherited retinal disorders, really the large unmet need is that we need to find treatment for patients to stop or repair blindness.

As you can see at top of the list here is Usher syndrome. You have heard a little about nonsense mutations today and that's a key focus, and on Usher syndrome. What causes Usher syndrome? As on the slide presented, it's an inherited disorder it passes from parents to a child through genes. And sometimes the genes are altered, or mutated, and where the problems occur, and in this case, affects the eye. What are genetic mutations? You have heard a little about that already today and I have had a

Scroll to top

nice introduction for what I'm speaking about. Genetic mutations are errors in our DNA code.

These are seen in the DNA, RNA and the protein. When you have errors in the DNA code, these cause our cells in the body to behave abnormally.
So these mutations or abnormal actions also affect the proteins in our body and that is where our focus is. Some mutations cause proteins to be shorter than usual, or sometimes that's referred to as being truncated and these shorter proteins don't work properly. Instead of being able to function, they're just broken down or degraded in your body, or in this case, in the eye.

Many patients have Usher syndrome due to the shorter or truncated proteins. In our case, we focus on full proteins or functional proteins and you have heard a little about that today and I will go into more detail about that and so our focus is very simple.

It's on restoring protein function. If you have a non-functioning protein or a mutated or shorter protein in your body, our drug, that we're proposing, is to work by this read-through process that makes mutated proteins become functional or full proteins.

Some of you may have heard about aminoglycosides or antibiotics, or the antibiotic called Gentamicin, so what we know about aminoglycoside or Gentamicin, promotes read-through that fixes mutated proteins. What we have done is take the antibiotic properties out of Gentamicin and really focus on the ability to do read-through and create the protein. Why would we do that if we know that aminoglycosides can do read-through and help restore vision in patients with Usher syndrome? That's because there are side effects with Gentamicin and if we take away antibiotic activity there and really focus on read-through activity, we could potentially have a drug that can do good things for your eye and not have the bad side effects so at Eloxx, we're developing specific glycosides, that are not aminoglycosides, but have a fancy term. Eukaryotic ribosomal selective glycosides, ERSGs and they are better tolerated and safer and can better restore protein in the eye and thus we feel it has the potential to be an Usher treatment.

Before we can go into patients, I know you're anxious for us to go into patient clinical trials, we need to make sure that potential drugs are safe. First way we do that. We look into rabbits. We do special testing in rabbits. May not be an eye exam but is a specific study, electroretinogram. And study it before we put it before we put it in patients. You can see the study, we did in rabbits, the ERSG and compare it to antibiotics because we know the antibiotic has the safety side effects and as you can see here in the study that we performed, compared to the antibiotic, our Eloxx drug did

Scroll to top

not negatively affect photoreceptors which is what the ERGs measure and the photoreceptors are the neurosensory part in the retina in the back of the eye that sense light and make vision.

As you can see on the slide here the normal box and the Eloxx drug box look very similar. ERG is not affected. Whereas if you used Gentamicin, the ERG is affected, not in a good way. That's the first step. Safety and tolerability. Does it work at all? We have an idea that it might work. The next step is looking at this in cells of the body that have an Usher syndrome mutation.

In this case we used USH2A and done this in other mutations and I will show you this one to help demonstrate that. So as we said, we're not focused on the DNA. We're focused on making functional full-length proteins so that's what we're going to look at. Increasing the full-length or functional protein produced in cells that can help in the case of Usher syndrome patients see better.

As you can see on the slide here and we demonstrated in more than one of the compounds that we're currently looking at to put into clinical trials, in the cells with the Usher mutation, we can in fact restore functional full-length protein which is really exciting.

We're hoping to go from cells and animals into patients with Usher syndrome and this is a really exciting time for us, and a first step.

So how do we get it there? Also very important because it's your eyes that it may be going into. And what we are proposing is not a surgical procedure. But actually delivering the drug to the back of the eye, what we call the vitreous cavity and drug is able to get to the back of the eye, the retinal pigment epithelium, both layers needed for functional vision. So that's the next question. We know in cells we get a reaction. In rabbits, we show safety. Can we get it into a bigger eye? And the back of the eye where it can actually work for an eye disease? And in fact we have put this again in rabbits into the vitreous cavity, and we have been able to show that we get good levels in the retina and pigment epithelium, two areas in the eye where the drug needs to go to get positive effects and restoring vision in patients with Usher syndrome. We are excited about the fact that we could potentially offer it as an office procedure. Some of you may be familiar with this procedure if you have family or friends with macular degeneration or diabetic eye disease, because this is the way that it's delivered in those patients as well. It's a procedure with much more safety, and the retina specialists are much more familiar with drug delivery in this sense.

Scroll to top

I hope to come back next year in a meeting to let you know how much we have progressed about potentially treating those of you with Usher syndrome. This is still in development and we are hopeful. I will share your passion today with this moving forward, in read-through for Usher syndrome, and helping to make functional protein which will hopefully prevent vision loss or improve visual loss and visual function in all of you.

We have shown safety and tolerability in doses that are much, much higher than we have planned to use in patients. We are excited about that. Much needed step about safety. Can't move on without that.
We have shown an effect, read-through. Better function in the back of the eye and we have done this in cells representative of Usher mutations and can get it to the back of the eye with the delivery that we would like to do, which we think is a better in-office procedure.

And more tolerable to patients and their families to get the drug into the retina where it needs to be and the fourth goal, the final goal, I wish I could have put it in yellow for you, because it's the yellow spot for us, really bringing a good treatment hopefully to patients with Usher syndrome so we can save and improve vision with patients going forward. I really want to thank you for your time. I am completely inspired by this moment today. I have never seen such passion. I look out and I see a lot of smiles, and people really excited about the future.

And we do have a booth right outside. I'm here all day. And I have gotten a chance to meet a few of you before this presentation today. I am looking forward to meeting everybody and share your excitement and yellowness. Thank you all very much. And have a wonderful day. [APPLAUSE].

>> Thank you Dr. Schneider. You will have lots of opportunities to see her. They have a table out there. We will take a break. We will take a half hour break and be back at a quarter to 11, and try to get ourselves a little bit back on schedule. That's fine.

Take a quick break. And we will be back in about a half hour or so. Please see Nancy if you haven't registered yet. Thanks.

[Break] >> Hi, if we can get everybody to take their seats and we can get started again. If we can get everybody to take their seats, we will get started in just a minute here. Just trying to get everybody organized. Okay. We're going to get started again. Thanks, everybody, and I hope that you are finding good connections here. I met a lot

Scroll to top

of people who are terrific. I just met Ching who is in Baltimore, from China. She is outside. I was going to connect her with Ben. I'll talk to her in a minute.

Our next speaker is Christine Kay, director of retinal genetics at Vitreoretinal Associates in Gainesville, Florida. She will talk more about what they're working on, so if you want to come up, Christine. It will take just a second to get her hooked up.

"RUSH2A Study - the Importance of Natural History Studies" Christine Kay, MD

>> Christine Kay: Hello, everyone. Again, my name is Christine Kay. It is a distinct honor to be here with you all today. I have been already significantly moved through some of the talks that we heard this morning, and I am very honored to have been invited as a speaker. The reason I'm invited as a speaker is because I'm one of the principle investigators in a clinical trial focusing on USH2A, a trial sponsored by the Foundation Fighting Blindness and it's a natural history study. I will talk about, at the end of my talk, what natural history studies are, why we need patients to be involved in them, and what they help develop as far as drug therapies and treatments. I'm in Gainesville, Florida, at a practice called Vitreoretinal Associates, previously professor at University of Florida working on AAG gene therapy, vector development, in a lab with Bill Hauswirth and I have a significant background in gene therapy as well.

These are my disclosures, which are primarily letting you know about the clinical trials that I am involved in, so we actually have seven ongoing inherited retinal dystrophy trials at my practice. One most relative to the discussion today is the RUSH2A. And that's the disclosure that I am receiving funding. I am involved in studies on Stargardt, pigmentosa, and other studies listed here.

Usher syndrome is what we're going to talk about today. The prevalence is difficult and controversial so my first slide here is a controversial number because it's very difficult to define the prevalence of Usher syndrome. More than you thought. Depending on what literature you search and what search base you use, you can get one out of 10 thousand or one out of 100 thousand. I think the prevalence lies in about the one out of 30 thousand range. Much more common than we used to think ten years ago and part of that is what Dr. Kenna told us earlier, hearing diagnosis with infants and improvements in genetic testing.

Scroll to top

Usher syndrome is the most common cause of hereditary combined deafness and blindness and think of Usher syndrome involving early onset hearing loss and there are some caveats there on how early it is, depending on the type of Usher we're talking about and the vision loss can either be called retinitis pigmentosa which is a common sort of "catch all" term or potentially more correctly, rod-cone dystrophy. The rods typically affected first, night vision, and peripheral field vision cells and the cones being our central vision cells.

When you see us say rod-cone dystrophy as opposed to cone rod dystrophy, what we mean by that, retina specialists, the rods are affected first and more progressively, and the cones are affected later, but to a lesser degree. This means the peripheral visual field is shrinking overtime, but the visual acuity, the reading vision is often spared forever with patients with this disease and that is good news and something important in the research field to ask, why does that happen? And how can we keep that central cone health as long as possible, if not forever?

This is an autosomal recessive disease and we have some genetic counselors in the room today I heard. I won't get too much into this. Involves a mutation you gained from your father and one that you gained from your mother. Typically your mother and your father would not be affected because they have a healthy unaffected copy of the gene. For every cell in our body, we have two copies of a gene body. For the protein in the cells required. And if we have two copies, both need to be mutated in order to have the condition.

This is an overview of the types of Usher and you have probably all seen and studied this and heard about this. Oh, am I not -- are they advancing for me? Because I have been forgetting to advance. I think I have been forgetting to advance. He is doing it for me. I have been forgetting to advance my slides but I think we have a very good IT crew doing it for me.

Type 1 is less common. Makes up about 40% of Usher syndrome. This involves more profound hearing loss. These are typically the babies or the infants that are being diagnosed. So when you hear the stories of seeing Dr. Kenna early in life in age 0 to 3, these might be patients diagnosed with Type 1 and tend to have vestibular. The part of our body that helps us with balance issues.

They may have difficulty with walking or late onset walking and that's an important question as a retina specialist, I ask patients, what age did your child walk? If they are 10 years old, child will not remember the answer to that question and mom could say they have delayed walking. On average, the child starts walking around age 1 but if they did not start until age 2, that can be a sign

Scroll to top

of vestibular or balance dysfunction. These are the genes -- I'm not going to read them out for the sake of our interpreter here. But you can see five genes associated with Type 1 and others identified but not the genetic mutations yet.

USH1B is important to point out, also MYO7A, it's the trial that is currently ongoing using a lengthy virus vector.

Type 2 is the most common. 60%. Less severe hearing loss and RP is milder and later onset. The patients do meet their walking milestones and usually walk by age 1 because they don't have the vestibular imbalance dysfunction. The most common you will hear is USH2A. We will hear about antisense oligonucleotide therapy and then translational read-through. Type three is most rare. Less than 3%. CLRN1, most common in Finland, it comprises 40% of the patients. This is a patient with USH2A. You can see classic findings. I don't know if I mentioned but I'm a retinal specialist and a retinal surgeon. I am going to show some pictures today and I hope they are instructive in some way.

I'm going to point out some highlights of what we find abnormal on the eye exam. One thing, the optic nerve can get a bit pale over time in patients with Usher. The other thing you see a yellow hue, rather than a red hue. You can see the retina is spared. Everywhere else outside that, the retina looks white or yellow with little black spots. We call the black spots bone spicules and you may have heard that word. That's what the RPE hyperplasia looks like and that's from the break up or build up of debris from receptors over time and we see that in the peripheral retina because the cells are affected first.

There are actually two -- and I know this is an Usher Syndrome Coalition, but there are two phenotypes, and we learned that word this morning. That involve USH2A, the way that a gene mutation manifests. The way we see it in a human. Usher syndrome Type 2A we're aware of, hearing loss and vision loss and there is non-syndromic USH2A in the category of RP and we call it RP39. These patients just have vision loss. Does have hearing loss. Even though we learned it's a ciliopathy gene affecting the hair cells. It should also affect the hair cells in the ear but it doesn't in RP39 and it just affects the patient's vision. And additionally there is a paper showing that the RP in these patients may be less severe.

Here is a picture from a paper. You can see the website on the bottom if you want to look. The top A and C show a patient who is normal for comparison. B and D which is on the right side, show a patient with non-syndromic Usher. Normal RP and look just like Usher patients because they have the bone spicules, waxy pallor or paleness and the atrophy of the retina.

Scroll to top

The bottom two pictures I'll get to in a minute called an OTC. Anybody in the room have their retina doctor show or take this picture? Raise your hand if you have seen one of those pictures before in the bottom. Excellent. We call it OTC. Optical coherence tomography. Basically an ultrasound picture. Not saved. You can do it as many times as you want.

It's an ultrasound wave we take in the back of the eye. Put a machine up to the eye, take a very quick picture and look at the thickness of the 250 micron thick tissue called the retina, at the back of the eye. And those are the two pictures on the bottom.

It's going to be hard to see if I can have a pointer. So the pointer here, this is again the normal patient. There is one layer here that's black, the outer nuclear layer. That's where all of the cell bodies live. We will get to an important layer, the line here called the ellipsoid zone and we need the outer layer to measure the function. And we can advance with that in clinical trials to see how many photoreceptor cells are remaining and how they progress over time. You can see the curvy picture on the bottom. The reason it's curvy is because this patient's eye is actually shaped a bit more curvy because they are nearsighted. The most important thing, you see that black area, it kind of goes away, versus from here to here it goes all the way across.

That would correlate to the patient having a 10 or 20 degree of visual field remaining because the motor receptors go from about here to about here as opposed to all the way across. That's a very important outcome measure we can follow in the clinical trials.

Speaking of clinical trials, RUSH2A is a natural history study and I think it's useful to look at clinicaltrials.gov website and be aware of NCT, the national clinical trial number. If a trial has gone through registration with clinicaltrials.gov, it will have a valid number and if it's a treatment trial, they will include FDA IND number. Investigational new drug number if it's a validated good clinical trial which has gone through FDA's approval to do the clinical trial. Does not mean they have FDA approval because you have to run the clinical trial first and see if the drug works and is safe and go through the approval process.

At the end of the day, we want for a drug to have gone through an FDA approved trial and submit for FDA approval which can take one year typically or sometimes do a six months expedited review.

For instance with the drug we heard today Luxturna, RP65, through Eloxx therapeutics, the FDA respects that they need to go a little faster when the trials

Scroll to top

are so important so sometimes we have expedited six-month reviews for diseases that are very rare and aggressive.

The sponsor of the -- I'm going to let them switch. The sponsor of our trial is actually the JAEB Center actually out of Florida and I think it's probably most important to point out that the originator of this trial is Foundation Fighting Blindness and the chair, Jacque Duncan, a good friend of mine, sits on the Foundation Fighting Blindness and raises funding doing research for these rare diseases.

Other collaborators were Duke and Oregon because they used them in the reading center to look at all of the images. There are many sites. We call this a multi center site study. Vitreoretinal Associates, that's mine. Emory. Wilmer. NEI. Duke. Columbia. We have the whole U.S. covered here. Toronto. Germany, Netherlands. To summarize, RUSH2A is to characterize the natural history of progression in patients with USH2A related to retinal degeneration associated with USH2A. And study objectives are to characterize the natural history over four years. It's a good time for the study.

Remember, they are time consuming. Expensive. The funding has to come from somewhere. So a four-year trial would be a good relatively long trial but it would be nice to follow patients for 20 years, but not realistic.

Some of the measures we are following are functional outcome measures. How are the patients actually seeing? We test the vision with very different methods. Static perimetry. And then microperimetry where we look at the part of light sensitivity with the anatomy with what the macula looks like. Full field stimulus threshold is a good test for patients because it does not require fixation. Patients basically sit in a dome, a bright light flashes and the patient says yes or no if they see that and we can quantify the light sensitivity the whole diffuse retina has, whether they can fixate or not. In some patients with poor vision, it can be difficult. Retinography, most in the room with Usher syndrome probably had at some point. Remember it not fondly. Electrodes on the eye. And today we use the filament, the little electrode rather than the contact electrode that sits on the eye.

This is an hour long test, lights flash, it's a relatively memorable test. If you had it as an infant, you were likely under anaesthesia to do it so you would not remember that. And visual acuity, you would remember having the eye chart in front of you so see what you can see and how far you can go. We look at how thick is the retina? The OT test. The ellipsoid zone or EZ. Want to be able to investigate structure, relationships and a lot can happen and progress in four

Scroll to top

years, depending on the patient. It's important to see. If patients are not improving, it's important of a finding as well. There are two cohorts. There is a longitudinal study where we follow the patients for all four years and patients have to meet criteria. Patients with vision of 54 letters which is approximately 20/80 on the Snellen eye chart. Have to have good fixation, do the visual eye test and have a visual field of greater than 10 degrees and if they have those, get enrolled into natural history longitudinal which is what you would think of where we follow you every year.

We did not want to exclude patients from the analysis if they had very low vision because we wanted to capture the disease, because often at a point, it stabilizes and is hard for the patients to do the test and these are for vision worse than 120/100 and most importantly visual field less than 10 degrees and enroll them in the cross sectional study which means that they are seen once. It's important that we see them one time. Very critical information and don't have to come back each year. The primary outcome measures are change in visual field, visual acuity and the OTC picture which I will show you pictures of. Also check the ERG and full field visual sensory threshold or the dome with the bright flash where either the patient sees it or not. Probably the most important measure we're going to be able to follow here is the EZ area on the OTZ and this photo right here I know you can't ahead. These tiny little lines are shown in the retina. The bottom is the layer of RPE cells, epithelium cells and one of the retina, the big fat line right there is the ellipsoid zone, tells us about the photoreceptor integrity. And if it is gone, likely don't have vision in that area. It can be hard to read these. We need a reading image. And secondary are baseline, cross sectional based on mutation. They go through extensive genetic testing. Mom and dad, confirm which mutation mom has, dad has. Ask about history, comorbidities and see if there association with other things we don't know about. Give them patient reported outcome questionnaires and becoming more and more important specifically for the FDA to approve some of the products. Want to hear that patients are subjectively having an easier time with their life and patient-reported outcomes are becoming very important in trials so looking at them in natural history studies is the most important and first time to evaluate them.

Inclusion criteria, patient has to be willing and able to participate in all four years. They can't be traveling extensively and unable to come for visits. They need to be over 8 years old, or above 8 and they have to have two mutations in the USH2A gene. From an ocular standpoint, both eyes must meet criteria. Have to have rod cone degeneration, have the ability to form perimetry reliably. If they have anything else, very rare, inherited retinal dystrophy gene, we can't include

Scroll to top

them of course because that would muddy the water of what we are looking at. We ask them specifically, do you expect to be enrolling in a treatment trial during the next year? It is very difficult. We can do it but don't like to take a patient out of a natural history study and put them in a treatment trial. We can do it. Of course if patients want to do that, but we lose patient's data at that point. We ask them if they expect to be and that, as you'll get with the next talk, it's sometimes happening because there is treatment going on with USH2A trials.

We ask if they have taken Plaquenil or other medications that can affect your retina for over a year. Can't have hemorrhage, vitreous retina, can't have had a cataract or glaucoma diagnosis or anything that would make sense to you with an eye problem, we don't want to muddy the water so we exclude the patients.

This is an example of a patient from my study. Of course I am allowed to share with you. You can see the power of the optic nerve. You can see the atrophy or the thinning of the retina which is the speckled whitish color and the thin vessels. The photoreceptors are dying and there does not need to be as much oxygen to supply them. The vessels get smaller.

This is a patient in my study. I'm going to show you the remaining ellipsoid zone in the patient. We scan up and down and you can get the area of the remaining ellipsoid zone and that's something we can quantify and study and that's very important as part of the RUSH2A trial. And this is the kinetic visual field. For the picture, we give them a button and click with the light stimulus. This patient did enroll in a longitudinal study because they had more than 10 degrees remaining on the visual field. And on this photo it's about -- 10 degrees is sort of in here.

This patient has more in the 40 degree range under the black which is the large light.

This is static visual field, another way to test where we put a light dot stimulus to them and this is the gray scale where it shows you the data very nicely. The area they can still see is red hot looking area and black is the area they have lost. Here you can see the picture of the central sparing and the peripheral loss, visual field. So in summary of the RUSH2A study, started in August 2017. Expect to complete in January 2023. If you look at clinicaltrials.gov, it says it is ongoing no longer recruiting. Great. 137 patients. 105 in the primary cohort and 22 in the secondary cross sectional cohort. I would recommend looking at the clinicaltrials.gov website if you would like to familiarize yourself and keep updated with the trial.

The last section which is very short is to go over the general importance of natural history studies. Why are natural history studies important? The

Scroll to top

information obtained from natural history studies can play a very important role in every stage of drug development.

Number one, in drug discovery, learn from natural history studies what the disease is doing and also learn very important genetic information and often we learn from the natural history study, new ideas to treat the disease and enables the discovery drug part of the development. And helps us, most importantly, design clinical trials. We learn which outcome measures are important? How stable are they? How reliable and repeatable are they? How difficult are they for a patient to do? We put the patient often through a day of testing and they tire and fatigue and it's difficult for them and for our clinical coordinators and staff who are doing the tests and we have to pick and choose who are the most important tests that we can pick into a reasonable seven or eight-hour day with a nice lunch break so patients can get through the tests.

We can't do every test we want. We want to support the marketing of a drug eventually and post-marketing and that's finding a patient. When we identify patients in natural history study, we now have a pool of patients or family networks that once a drug is available, we can recruit patients for treatment.

Uses of the natural history, again, is identifying the patient population. Identifying outcome measures. Identifying biomarkers which are different ways to evaluate in the human body to see what it's doing and how treatment might be affecting it, and the design of external controlled studies.

There are several types of natural history. The easiest to think about is retrospective versus prospective. Retrospective, we look at previous data and gather chart reviews and look at the history of the natural disease. We can do that over a longer period of time and in fact has been done with multiple diseases. However, the date is very muddy. I have been using that word a lot today. Different sites have used different measures. Different machines. Nothing is controlled and reliable, versus a prospect of studies. We know exactly what tests we're going to use, which machines are used and how often we're going to use the test. Everything is perfect as far as the data we require from a prospective study and we can select patients specifically for that trial.

Considerations, natural history are non-invasive. We're not going to treat the eye. These patients can go into a future treatment trial. Patients always ask me that question. Of course if you have been in a natural history study, never an exclusion from being in a future treatment trial. And it is not the case if you have been in a treatment trial and can still get into a treatment trial.

Scroll to top

In fact the opposite is true. Let's say you have been in a treatment trial and had surgery and stem cells placed under the retina or cells, you may likely not be a candidate for a future clinical trial. Think about it. If the next company wanted to come along and evaluate if the therapy is working, they need to know it's their therapy and not the one you received five years ago that is helping you. And so usually you are excluded from future trials if you have been in one surgical trial.

Oral drugs are different. Usually a wash out period is allowed. If you have been in Akouos trial where you have taken an oral drug or several trials after that, gene trials would historically allow a patient taking an oral drug, to take three months off of the drug and then go into future therapy gene trial.

Back to natural history trials, you're good to go. You can still be any treatment trial and in fact you will likely be highly recruited for a treatment trial because you have excellent data obtained on you. We already know you are a great candidate and we have your information and can find you. That's probably the easiest way to access you, get into a natural history study if possible. These are not very time intensive. Once a year tests for the patients. They are camped out for a day with us but then we see them in a year. It replaces my clinic visit with them. I don't need to see them two days later to see the same thing. Trial visit is the once a year visit with me. It's true, you can withdraw. We don't like that. It's difficult for us because we lose the data if you go into a treatment trial.

This is happening right now. And you will hear the next talk on ProQR. If you have a certain Exon, 13, you can be recruitable. Patients in the USH2A trial might have the exon and it's up to the patients to make the decision of when to withdraw and go into the treatment trial. It can absolutely be done. You're not stuck.

Ideally we would like to keep the natural history patients, if possible, in the natural history trials. Some examples of some natural history trials relatively well-known and really paved the way for future gene therapy. PROGSTAR, Stargardt disease, it was a huge trial done for patients with Stargardt disease. Data used by Ophthotech for an inhibitor.

AGTC, I'm with their trial, first they looked at achromatopsia, they did a several year natural history trial and then did their own treatment trial and we, because I was investigating with that, we learned which outcome measures are good to inform us of how to run the treatment trial. And the RUSH2A trial, we will definitely be using this data to help inform some trials of outcome measures, how stable the patients are, or how they are progressing overtime.

Scroll to top

So what can I do as a patient?

Stay informed. You're in the right place. This is a wonderful conference. You have a wonderful network here of friends, families, educators, researchers and physicians to help you be aware of what's going on. Foundations like the Usher Syndrome Coalition and Foundation Fighting Blindness and Retina Foundation and multiple other nonprofits raising money for diseases for research and developments in the field. Attending conferences and seminars. You're doing that right now.

Website clinicaltrials.gov, I would always be aware of that. You can find some unethical trials on clinicaltrials.gov. I always tell patients if a trial asks you to pay 20 thousand dollars to be involved in it, I would steer clear of it.

Well-run clinical ethical trial typically would fund most everything in the trial, if not at least all of the testing, the treatment, and potentially even the travel so it's a significant red flag if a trial is asking for a significant financial investment from a patient.

And that is an important point. You can find some unethical trials on the clinicaltrials.gov website. It's important to talk to a doctor or inherited retinal dystrophy specialist. That would be best.

There are few of us. Not many retina specialists in the country who specialize. But we are geographically distributed. Down in Miami, there is an expert. I'm covering the top half of Florida, Georgia and South Carolina. I sometimes see patients but there are inherited retina dystrophy experts across the country, people who are aware and interested in this. Try to find someone who has awareness in Usher syndrome or retinal dystrophies. If you don't have the data yet, this is the day to get it. We're in a fast moving field. We have multiple ways to get free genetic testing outside of insurance coverage including Foundation Finding Blindness, Spark has a panel that is free on their website, as of two weeks ago.

There are multiple ways to get genetic testing done and I would highly suggest getting genetic testing if not confirmed yet.

And lastly, if a natural history trial is available to you, get involved. It's very important to the field. We appreciate the patients that are involved in the treatment trials.

It's how we are moving the field forward. It's a huge investment of the patients' family and time the clinician, and in the end, the person that you're helping is you,

Scroll to top

and your family and your children because this is the only way we can move this field forward.

It's inspiring to be here today. I left my three little girls at home with daddy in Florida. They are going to ballet right now with daddy. Hopefully he made it there with them. This is my family. Hopefully you enjoyed my presentation today and I will be at the lunch if you have any questions.

[APPLAUSE].

>> Mark Dunning: So before I want to introduce our next speaker, I want to take a couple of minutes to make sure that everybody has context around what you have heard so far today. The first thing is that you're going to be able to ask questions to the researchers, and as you can see, they are all very impressive and very interested in connecting with you guys.

We're going to have a panel after this. You can catch them then, also at lunch and on the breaks. They are more than happy to talk to you. The other thing I wanted to tell you.

This morning we talked quite a bit about Usher 2A. I will talk more about it in a second. When I stood up here five years ago, very little was being done on Usher 2A and it's the most common form of Usher syndrome. The fact that there was nothing at the time was extremely disappointing to the community and now we are spending the entire morning talking about this and what you're seeing from the Usher 2A side of things, you are hearing about potential treatments that are coming from Dr. Schneider.

You heard we have an Usher natural history study being done on this. You understand how the disease works and that's extremely important when we talk about getting into treatments. We need to know in clinical trials, does this treatment actually work the way we think it will? Not just in rabbits as Dr. Schneider talked about, but the only way you can do that is if you have natural history studies about the way the disease works.

We have come an enormous way with Usher 2A and over the next few years, we're going to be standing up here talking about all different types of Usher syndrome. 2A makes the most sense because it is the most common cause and it's great to see how much has changed over the last few years here. Before I get to the next speaker, I'm going to take two minutes. I know the interpreters asked for a minute and we need to get everybody hooked up. So if you give me a minute, we will move on to the next speaker. [APPLAUSE] . >> Okay, so we are ready for the next talk. We have gotten the interpreters reluctant to come up

Scroll to top

here because they know they are going to have to spell oligonucleotide over and over again.

Our next speaker is Dr. Aniz Girach from ProQR and he is going to talk about a treatment being developed for USH2A and I will let him say oligonucleotide again. Welcome. [APPLAUSE].

Scroll to top

“Qr-421a, an Antisense Oligonucleotide for the Treatment of Retinitis Pigmentosa Due to USH2A Exon 13 Mutations” Aniz Girach, MD

>> Aniz Girach: Thank you, Mark and thank you to the organizers for giving us the opportunity to speak here today. I'm an ophthalmologist retinal surgeon and I am the chief medical officer for ProQR Therapeutics. A company based out of the Netherlands and we also have a base in Boston. Cambridge, Massachusetts. I live in the UK. I came from London here. And it's my absolute pleasure to be here with you all and share the day with you.

I'll be talking to you about our latest treatment, investigative treatment still in human study. QR421-a and it's an antisense oligonucleotide. Sorry. We will show the ASOs. Hopefully that will help everyone. ASO stands for antisense oligonucleotide and we are looking at USH2A due to a specific mutation and this was mentioned earlier by Dr. Kay, exon 13 and so ProQR is a publicly-funded company and I am obliged to show you these forward-looking statements.

I feel as an ophthalmologist, may be bias here, the eye is the ideal target for antisense oligonucleotides. You don't need large doses of treatment in there and can help in terms of safety treatment of the eye. Safety and side effects and also relatively immune privileged site. When introduced into the human body, recognizes those as foreign and amounts an immune response against those and that happens often in the rest of the body and sometimes neutralizes the treatment.

The eye is slightly different and we have seen from literature and research that is done that the immune response that is mounted against any treatment in the eye is perhaps not as advanced or profound as it is with peripheral treatment issues, and it can work better in the eye. Not always. But it's a good chance to utilize that fact and look for treatments in the eye. And the next reason for choosing the eye is because you can look into the eye. When you introduce treatment into the eye, you can see where it goes.

And often like Dr. Kay mentioned in the previous talk, can use various instruments and visual techniques such as OCT, and visualize what is going on in the back of the eye in order to see what the treatment effect is in the back of the eye. That is a great advancement over treatments in the rest of the body where you can't see what is happening.

And you can't visualize what is actually going on, whether the treatment is actually working or not. And lastly, the eye has many reasons. Because there are over 300

Scroll to top

genetic causes of blindness and many of these have been mapped to specific genes that we are already aware of.
And therefore, there is ample opportunity for us to be able to target the eye in order to look and treat, potentially treat causes of blindness that are there. And this has been touched on already with previous speakers. The way that we see with the eye, the light comes in through the front of the eye and goes straight through the middle part of the eye, in the lens, and through the back of the eye into the retina. Retina is the sensitive part of the back of the eye and it covers the area all the way around.

It allows you to see clearly at the end of the day. What the light does is it goes through some chemical transformation into electrical signals and does that in the retina and the most sensitive part of the retina and that's where the photoreceptors live. You heard about photoreceptors and there are two types. Rod and cone photoreceptors and these play important parts in Usher syndrome and many other diseases of the retina.

The light is therefore converted into electrical signals and those electrical signals are then fed back through the optic nerve and into the brain, into the visual part of the brain, in order to be interpreted. And that's how we see.

Now unfortunately as you may well be aware, with patients with Usher syndrome, there is a defect and genetic mutation that causes a break down of that process occurring. And we know that there are lots of other diseases which regroup as an umbrella term called inherited retinal diseases, which really are the cause, many of the causes of blindness today.

But I believe that the management of inherited retinal diseases is really coming of age.

The reason for that, increased awareness, meetings like this. And it's great to see so many people here with a packed house, ultimately to be able to share with what is going on in the Ushers community. And also you have heard, there are various ways in which diagnosis is helping us move forward.

With moving forward with accessibility and moving forward, it's helping to pave the way with the kind of mutations that are there, and leading the blindness, and therefore we can better target diseases with the appropriate therapeutic treatments moving forward.

We have already heard about treatment options out there. Gene therapy and in particular you heard about Luxturna. Luxturna was the first gene therapy approved for inherited retinal disease, IRD in the eye space.

Scroll to top

It was for a different disease called LCA2. And the good thing for that, the good thing for all of us, as we think about other eye diseases that cause blindness, the Luxturna model has paved the way forward for us to look at diseases in a similar way and open the floodgates for companies that want to explore treatments in the space. And that's good for people like me as an ophthalmologist, and good for people like you, as patients as well.

And a whole lot of other gene therapies have been looked at as well. And we have heard about some of those in the previous talks. The focus of my talk will be more on RNA therapies and ASO. The ones that I will talk about have been approved for over diseases but of course to highlight, the treatment that we're looking at is an investigational treatment and it has not been approved yet. I will stress that over and over again. I want to actually clarify for people the difference between RNA therapeutics, ASO and gene therapies.

Often people get those confused. We know that ASOs are very specific to a target. So are gene therapies as well. The advantage of gene therapy is that you have one treatment, and that one treatment usually is enough. With ASOs, you often have to retreat. You will hear more about that later on in my talk. With therapy, you use viral vectors, may have heard of adenoviral effective viruses, and these transport the genes themselves and deliver the payload into the eye and in the ASO you can use the treatment, direct it straight into the eye and it's a relatively simple injection, the vitreous cavity, which we heard about before whereas most gene therapies require complicated therapies, general anaesthesia where the gene lies.

The advantage of ASO over gene therapy, is the current technology with gene therapy does not allow for treatment earlier in the disease because of the amount of gene therapy you put into the eye, and potential safety and toxicity issues that arises as a result of that. Often with gene therapy, you have to wait until the end stage of the disease, and try to protect and preserve that central vision that is left there at the end stage of the particular disease that you're targeting.

And like gene therapy, ASOs, because they're injected into the vitreous cavity, they have access to the whole of the retina and therefore can potentially target earlier in the disease with ASOs, compared to gene therapy.

So with that in mind, ASOs are typically introduced into the eye through the vitreous cavity, in an in-office procedure. And has a safety profile, and something that we're going to carry on in our profile. And usually ASOs have a broad distribution throughout

Scroll to top

the entire retina, and have a long half life and do not need to treat with ASOs as frequently.

In our case, you will see later on, it's a twice yearly treatment. There are over 300 genes causing inherited diseases with retinal blindness and over 50 mutations of genes that lead to many thousands of targets for companies. In ProQR we believe that the ASO technology allows us to get up to 25% access to many of those mutations or disease-causing defects that are there.

But with specific constraints of safety and other things in mind, we believe that without technology, we would be able to target over 100 of these mutations ultimately. And we're not the only ones working in this area. There are many other companies who are excitedly working in this area. And that's of course good for all of you, and good for all of us in humanity.

Here is a very busy slide of the ProQR development pipeline. I want to stress here none of our drugs are approved. They are still going through clinical trials or in research setting of some sort. Our front-running program is a drug called Sepofarsen for a disease called QR-110. I will talk more about the human study, QR-421-a. However, I want to mention that we have a number of other programs going on, including two other programs in Usher syndrome coming up, which are currently in the research setting. Which I am hopeful, if all goes well within the company and the research being done, over the next few years, we will see the drugs get into human trials, in addition to the ones we have at the moment.

With the excitement going on in the whole field and with other companies working in this area, as aggressively as we are at ProQR, I really believe this is a good time for all of us, and actually a very exciting time because I believe there will be a therapy hopefully around the corner in the not too distant future for us.

So I want to start my talk talking about a disease not related to Usher syndrome, Leber's Congenital Amaurosis Type 10. I will shorten it to LCA-10. Like Usher syndrome, there is a mutation and problem with the back of the eye, which is very similar to the type of mutation we're going to be looking at in Usher syndrome. And we have already done Phase 1 and Phase 2, early human trials in this disease.

And we actually have very encouraging results that I want to be able to share with you and like I said, the similarities between this treatment and the disease, between Usher syndrome and the type of treatment we're looking for with Usher syndrome, is

Scroll to top

immense. And we want to be able to translate what we have seen already in the Sepofarsen LCA10 scenario, into the Usher syndrome scenario as well.

Here is a set of data, first human study of LCA10, Sepofarsen. I will not go through this in detail but in three patients, there is enough encouraging data, to go to the FDA to ask for an expedited review to move the whole platform forward into The Phase 3 setting which we have done so.
We were so excited about this data because a number of different outcome measures or end points moved in the same different positive direction that we intend for them to go into including visual acuity.

For example the FDA states the clinically significant threshold for improvement in visual acuity is by three lines on a visual acuity chart.

And for us, in this trial, remember it's only three months later in the early stages, we found that actually patients improved on a mean average of up to seven lines of improvement on the visual acuity chart. So this was really very significant and very exciting for us to see and this was coupled with significant improvements in other end points as well. And like I said, it really filled us with a lot of enthusiasm to be able to expedite this particular study and move it through to Phase 3. Why do I talk about that?

Like I said, there are a lot of similarities between that study, that treatment and the disease, to Usher syndrome and what we are looking at here. Both of these diseases are known as ciliopathies. You heard earlier the SEP290 protein are the Usher protein, the defect that we're looking at here in the Usher syndrome. The same part in the photoreceptor, as the connecting cilia. They have two connecting functions. Not exact of course.

And we know in previous studies, we know that ProQR has cell models that provide drugs in the research process. We take a small skin biopsy from a patient who suffers from a disease and then take those cells and grow them in a dish and take the stem cells and turn them into actual retina itself.

And therefore in the dish, we have a singular line, which is made up of disease retina of the patient that we actually had collected that sample from. And that allows us to be able to explore various treatments, drugs, chemicals with that cell line to see which is probably going to be best to take forward in development. Which drug is going to cause too much in the way of side effects and which drugs will not be suitable moving forward and this model is being performed in new ways, but we have a well-oiled

Scroll to top

machine in areas in order to fast track that process which can take many years typically.

It's include that mechanism, that drugs have come through, today, our pipeline. One works on the rod receptors, the area, and the other works on the cone. And QR41 which is for Usher syndrome has some preclinical data on animal models to support it. The first program on LCA10 did not have that and now has human data, that I referred to on the previous slide with the data that was so exciting.

So the combination of those two, really fills us with a lot of enthusiasm. The similarities between those two drugs and those two diseases, is going to be hopefully producing similar results ultimately in the human trial for Usher syndrome.

And the basic principles behind the treatment here are outlined on the slide. In the normal eye retina, there are photoreceptors there. In Usher syndrome, it's rod, the night vision photoreceptors that deteriorate first and we find in Usher syndrome patients, the outer segments of that photoreceptor are not formed very clearly or very accurately because the connecting syndrome where Usher is predominantly used is not adequately functioning.

Usher syndrome does not perform well and therefore patients like yourself, over a period of time, have a loss of vision because the photoreceptors are not functioning well. However, if the cell bodies of the photoreceptors are performing well, we can look at the scan that Dr. Kay showed you earlier, we can pick up patients who have restoration of the photoreceptors with treatment ultimately. That's the goal of course. The idea is if you have a restoration of the photoreceptors, you would then have functional vision coming again and the idea is that the functional vision allows you to hopefully either keep what functional vision you have, or indeed in some cases, as seen in an earlier program, having some element of restoration of some vision as well. And it is a similar scenario for LCA10 and that's what we have seen in the first of the human trials already there.

As you may know with Usher syndrome, disease progresses and starts typically first in infancy with hearing loss and then leads to night vision problems over a number of decades.

And of course it leads to peripheral vision loss, and leads to sensory vision loss as well as patients get into the 40s, 50s and 60s, depending on the severity of the disease.

Scroll to top

So QR421 is ASO is the investigative drug that we are looking at. What we have on the slide is what you need to know about the drug itself and where we are in terms of the process. Looking at Usher 2A syndrome due to exon 13 mutation. That's very important. We believe there are about 16,000 patients but I would think differently, seeing what we have here. Be part of registries. Get yourself involved in that. Even if you're not interested in clinical trials, it's important to know accurate data of how many patients are out there and how many patients can potentially be treated ultimately.

This trial which is the first in human trial, was enabled by the Foundation Fighting Blindness and I want to recognize them for the financial support that they gave us in order to have the trial going forward. It has already gotten Orphan Drug designation by the FDA and fast track designation by the FDA and the first patient has already been dosed earlier this year in March.

All together we have four sites in the U.S. and two sites in Europe. The four sites in U.S. are in Boston, in Michigan, Portland - Casey Eye Institute, and in Texas as well. And the two sites in Europe are in Belgium and also in France.

So of course if you're interested in being part of any of the trials, there is eligibility criteria that you have to fulfill and it's important to be seen by your local ophthalmologist or through an organization such as this, to be referred to the sites. So a careful evaluation can be made on whether you would be eligible for the trials or not.

The basic principle behind the treatment itself has already been talked about before. Ultimately QR-421, the ASO, produces a shortened version of Usher but we believe that's actually functional already.

And the reason for that is highlighted on this slide. A complex data slide. The message behind the slide says with QR-421 treatment, there may be significant normal mRNA expression which can lead to restoration. And in order to validate that, we then continue that in zebrafish. Essentially the message there is with QR-421 treatment, appearance of the retina was almost back to normal and on the right-hand side with ERG assessments, the function of the zebrafish mutant retina was almost back to normal. That filled us with a lot of enthusiasm to be able to progress this treatment into trials.

Additionally we know from the data slides, that the drugs QR-421 lasts in the eye for quite a long time. At least six months after an injection. First in the human trial, we're looking at twice yearly treatments. The phase 1, Phase 2 is called STELLAR. Looking at patients, double-masked study which means that you won't be aware of the

Scroll to top

treatment, neither will the physician. It's randomized. You can have the treatment or get a sham controlled placebo injection which mimics the whole procedure in the eye.

That's quite a well-recognized method in ensuring in clinical trials, that the authenticity of the trial is up to the standards of the FDA.

The first goal is safety. We want to make sure that treatment is safe for patients and we can identify the right dose to move forward into further human trials. It's a 24-month study and we want to include between 18 to 30 patients in total. These will typically be adult patients. 18 years and older will be included in this disease. And I will talk to you a little more about the severity of the disease also. However, one of the key inclusion criteria is that patients need to have greater than or equal to 10 degrees of visual field still left in the active eye, A-study eye as we call it. At least one eye. In addition to that, patients need to have visual acuity of 20/32 or worse. The two criteria are important.

There is an uneven balanced allocation ratio between treatment and sham controlled. In favor of two to one. If you put yourself potentially forward, if a patient puts themselves forward for the trial, they have a two-times equivalent chance of getting active treatment as compared to a sham controlled.

And the reason why we do that is really guided by the FDA in order for us to generate a lot of treatment data to see if the drug is indeed safe and efficacious.

I will be talking a little bit more about the trial design and essentially some of the end points that you heard Dr. Kay talk about earlier, in order to really prepare you, not so much for this particular trial, although we would really hope that you would want to be part of this trial, but also more importantly, for subsequent trials to come in this disease space, in any case, that we hope you would be part of, whether it's part of ProQR's trial, or indeed any other company's trial or any institution-type trials.

The idea and hope is that after the first human trial, would progress into subsequent Phase 3 trial to get hopefully the drug approved for patients.

The first patient was dosed in March. Again, I want to highlight that. The study pinpoints, I can see more clearly here we're looking at patients whose visual field is really looking in the central zone area of having greater than 10 degrees of visual field, but actually having visual acuity of 20/32 or worse. For the sake of this particular trial population or this trial itself, we have characterized patients into either moderate disease or severe disease.

Scroll to top

Moderate disease would be patients who have slightly more visual field that's there than 20 degrees. Severe patients would be those patients who would have slightly less than 20 degrees of vision but more than 10 degrees which is the minimum standard needed.

I'm going to talk about some of the end points, so you're aware of the kind of end points we think about in companies and it's not a bad idea to get familiar with those as we think through this.

The first one is full field stimulus threshold or FST. Has been used before in previous trials and as mentioned by the previous speaker, an improved way of looking at sensitivity of the eye, due to a flash of light, the flashing into the patient's eyes. It does not need any fixation, and that's the advantage. Of FST. Typically tests the most sensitive part of the retina.
It can pick up rod disease, rod photoreceptor disease or cone photoreceptor disease. And we want to see the treatment group.

And the second set of endpoints I am going to talk about is perimetry. It's how actively you can see, visual fields that you can see. There are active tools that we use to measure visual fields. Some of you are familiar with static visual field measurements where you put your chin into a dome and you have lights flash with different density in different parts of your vision and for you to press a button when you see that light itself being flashed.

The tool, the instrument itself is called the octopus which is the gold-standard tool we tend to use here.

There is a newer tool called the Medmont used for diseases like Usher's and I believe you're using it in the RUSH2A trial. And something that you're going to be using in our trial and hopefully much more sensitive to the types of disease like Usher syndrome, Type 2A and again, the idea is that we would like to see improvement. At least retinal improvement in the vision. And somebody mentioned earlier, micro perimetry. We are using the Maia device. That looks at the visual field, the 10 to 20 degrees you may have preserved. This is the instrument. You can see there on the right. And the goal is that we would like to see improvement. And there are various different ways to do that and various different analyses we can use. We will of course be focusing on visual acuity as well. Measured in a couple of different ways. When you and I go to optometrist, the Snellen has letters on the chart and more as you go down the chart, typically used in clinical practice but not a good sensitive tool to use in research. In the

Scroll to top

research we tend to use the chart on the right-hand side, the ETDRS chart. And it's useful because it has the same number of letters as you go down. And it's helpful when we look at trials and investigative treatments.

Dr. Kay already mentioned about OCT, optical coherence tomography, and this view is looking at the back of the eye, the back of the retina. And on the picture, the vitreous cavity of the eye is in the black space up at the top. The brain would be further down right at the chart to orientate you and therefore taking a cross sectional view of the retina.

You can see the layers and this easy line is of particular importance as we think about treatment because this affects the photoreceptors that are there and it's the primary problem in Usher syndrome, and others that affect pigmentosa, it's a very important measure for me as a researcher, FDA, and for you as patients as well. This is an important end point to look at. In summary, last slide, eye is the ideal organ for ASO and many other treatments. ASO treatment characteristics are ideally suited for treating eye diseases because of simplicity, ASO into the eye with injections and twice a year treatment. The half life.

There are many inherited retinal diseases. I spoke to you about the Phase 1/2 trial for LCA10 and Type 2A, exon 13 mutation and another, autosomal dominant, which is to start at the end of this year. There are many different end points and instruments. And this is good news for all of us because we can show whether the drugs improve outcomes for patients or not.

And there are importantly many companies working on these trials and many great researchers out there working with us and collaboratively. I think it's a very exciting time to be a researcher in the field in Usher syndrome, investigating in the area as well, and for you, as patients.

Because I do feel this is an exciting opportunity for all of us to ultimately find a treatment in the not too distant future. With that, I thank you for your listening.

[APPLAUSE]

>> Mark Dunning: So the next thing we're going to do here I have asked all of the researchers and clinicians to come back up front here and they are available for your questions, the next 45 minutes or so. I have a mic that I will run around to grab anybody that has a question and I ask that you raise your hand and Krista is here which is usually telling me to do something. So if you can just give us a minute or two.

Scroll to top

We have this Roger pen up here connected Roger pen that makes it difficult for us to have a panel. But we had a volunteer give us their Roger pen so we can use it with the panel.

We will get set up and then take your questions.

So just another minute or so, and we will have this started. If you do have questions, what I will ask you to do is just raise your hand, and I will run this microphone that I am holding here over to you. And we will get that question that way. And I will ask the panel to repeat that question, since they will have the Roger pen up here.

Here comes Shanna.

Okay, at this point I will hand it over to Marly Kenna who will moderate this. And I will hand a mic over to you.

>> Mark Dunning: I think what we will do -- I have a couple of announcements. One is about lunch. We're going to stay here until about 1:00. Get everybody's questions in and lunch will be right through the door, to my left. Your right. It's a boxed lunch. And once you're out there to the right around to Salons A and B, there will be places to sit. And I assume people can come back and sit in here as well.

And then we will reconvene at 2:00 for the afternoon session.

Scroll to top

USH RESEARCH PANEL Q&A

>> Marly Kenna: So I think right now, we're open to questions from the audience for anybody who is up here. I think once again, we are glad to be here. And learning as much from you as you are from us.

So I just want to thank you.

>> Yeah, this is a question for Dr. Kay. You mentioned -- oh, hello, hi everyone.

You mentioned in some of the research trials that the recruitment is already closed. Is there ever a time when some of the folks that they may fall off and you will open up recruitment? Or once it's closed it's closed and you can't start a new person?

>> Christine Kay: It's a good question. Specifically for RUSH2A, I can answer that, enrollment is completed. So even if someone drops off, we don't enroll new people. That's primarily because we want to complete the trial in the estimated amount of time. The 2023 estimated time. We occasionally will lose a couple of people. Every trial expects to lose a few patients over time. Whether it be natural history or treatment trial.

We write that up and let the IRB know the patient has dropped out and we don't typically reopen enrollment.

There are lots of caveats to that. Sometimes a second cohort will be done. For instance I'm doing the Akouos trial and we have the Stargardt measure and can be a Phase 2. A lot of trials expand into a cohort measure and it's not reopening the same trial exactly.

>> Mark Dunning: I see questions back here. The big room. Bear with me as I walk back. If I can remind the panel, if you can just repeat the questions.

>> Hello, this question is for the doctor from ProQR Therapeutics. You're saying that possibly there would be two injections a year into the vitreous. How do you expect the eye would hold up if this was like a yearly thing? If it would cause any scarring? Or cause any other issues to the eye itself?

>> Aniz Girach: So let me just paraphrase the question and let me know if I am accurately predicting that. You want to know with the twice a year injections, what type

Scroll to top

of side effects will be with the drugs? I would also like Dr. Kay, who is an expert researcher in this area, to comment on that.
The idea here the first in human trial, that's exactly what we want to try and look at. Which is, what are the side effects going on in the eye? We note that the side effects can be broadly lumped into two buckets. First, side effects into the eye itself. Remember, you can be all located to the sham control. But patients can get, historically at least, infections, hemorrhages can occur and those are known historically with any injections. We know that working with many trials.

And another specific that we're looking at in particular with these drugs, the ASO drugs, we want to try to find out what side effects the ASOs have within the eye. We will be doing a whole battery of tests including physical examination of the eye and many of the end points that you heard me talk about, to try to figure out what potential benefit of the drug could be. More importantly, what are the harmful side effects of the drug. And that would help inform which particular dose level we would like to then move forward into the next phase of the trials ultimately. By doing a summation at the end of the day, analysis at the end of the day, of all of the benefits, versus the risks that are there. If for example you put yourself forward to one of the trials and go to one of the centers and you are eligible for one of those trials, the doctor that will be there, principle investigator will go through with you, the more specific details in an informed consent, to make you aware of all of the pros and cons of entering into the trial. That will certainly be more detailed.

>> Christine Kay: I am a retinal specialist and retinal surgeon as well. To answer the question, are there specific risks of the mechanical intravitreal over time? I specifically inject patients for macular degeneration or diabetic retinopathy. Patients are coming in every few weeks. It's a 10-minute injection. Priming and numbing the eye. The injection takes a second, but the patients are getting the injections over a number of years and they are getting injections for different reasons and that does not speak to what Aniz was saying but from the mechanical injections, we have relative data that it's safe in lifetime, and even patients who have monthly injections.

>> I had a question about ProQR sites. So for the patients who are assigned to the sham procedure, if it turns out that the other patients are doing very well, will the patients in the sham procedure be eligible for treatment?

>> Aniz Girach: Yeah, so the trial duration is two years. The end points, we will be looking at, at the end of the 12 month time. And it will be a follow-up for patients who already have injection of ASO but importantly, for those patients who have the sham

Scroll to top

controlled one, if the external data monitoring committee believes that the benefit risk is positive, we can swap those and cross that patient over into treatments.

So it's by no means guaranteed. I want to stress that. But it depends on if the drug is working and the risk but it allows all patients to get access to the treatment.
>> Other questions?

>> Okay.

>> Hello. This is for Dr. Girach from ProQR. One of your slides said your technology can only target about 25% of new patients. And clearly a lot of us don't have USH2A and it seems like this is where the research is.

So my initial question was, who extrapolated some of the technology to some of the other genes are affected and your slide had said about 25%, so I am curious about that.

>> Aniz Girach: Yes, I think the question, I'll repeat it again, is that we're looking at USH2A due to an exon 13 mutation specifically. And of course there are lots of other mutations there that can cause Usher syndrome. The idea of these therapies, especially ASO therapies and gene therapies for that matter, is that you really have to target mutations specifically. The good news I think here is that although treatments such as this one, which are potentially targeting exon 13 are leading the way.

There are lots of other potential treatments coming along with companies as well. It's only a matter of time before hopefully, you know, many other mutation kinds are targeted. For example you saw in the pipeline slide, we're looking at different Usher syndrome patients. Not ready for the prime time human studies yet, but many other companies are looking for mutations as well.

And I think Dr. Kay, you had information on the slide as well about other mutations that other companies and other researchers are also targeting.

>> Other questions?

>> Lots of questions.

>> Yeah, I was curious about the various trials. At what page -- age, what is the earliest age you are admitting people to trial? I ask, I have a grandson with 1D, 20

Scroll to top

months old, already had cochlear implants. I am curious as to when you begin collecting baseline data.

>> Christine Kay: I can start. The question was specifically for Myosin7A and other forms of Usher, what is the typical age to start recruiting patients for an optimal trial or clinical trial. Clinicaltrials.gov is my cheat sheet because I can't recall off the top of my head with USH stat. There are different ages and trials but for treatment trials, we typically start with adults. Usually 18 and adults.
There can be pediatric cohorts. Luxturna for instance allows treatment up to age 4. I believe they had a 4 or 5-year-old in the trial and now the drug is approved for everybody, as long as it is not an infant where the eye is too small.

For natural history trial I was speaking about RUSH2A, our age of recruitment is 8 and older. We selected that predominantly because we want patients to be at an age where they can reliably do a visual field. Who has done a visual field in the room? Raise your hand. It can be tough. Can take 45 minutes or longer. You need some attention and the attention span of a 4-year-old or 5-year-old is not that of an 8-year-old. We select that age because we need reliability on these testing measures.

So it really depends on the trial. Depends on if it's observational or treatment. And the easy way to check is to look at clinicaltrials.gov because it will be listed almost all the time on the inclusion or exclusion criteria, what the age of recruitment is for a given trial.

>> Can I just make one comment? I think in terms of thinking about age, it has a lot to do -- whatever the disease is, severity of the disease as well. If you have a very mild version, in this case, Usher syndrome and have very usable hearing or very usable vision, people would be reluctant to intervene whether the patient is doing very well.

Most of the time, children are doing pretty well, but it depends a lot, in the Luxturna trial, the children are already severely affected. Good question. Mark, where did you go? Yeah.

>> Hello, this question is from Dr. Jeffrey Bohrman for the panel, anyone who can answer. For those who have severe retinal damage and we have given no hope because of the message we're getting from our doctors and the central vision area, are any of these therapies or treatments able to benefit people like us to restore our vision?

>> The question was, for patients with severe vision loss and severe retinal degeneration, are there any potential treatment options at that stage of the disease?

Scroll to top

Because it seems that some of the therapies we are talking about, we're talking about treating at an earlier stage. I believe that was the rephrase of the question. The two trials that I would mention, actually not specifically for Usher but specifically for retinitis pigmentosa would be jCyte, and I don't know if we already mentioned it, Reneuron is a trial that I would mention. It is for those with retinitis pigmentosa. It's not for patients with Usher syndrome, but jCyte involves cells where we do an intravitreal injections and the trial is ongoing right now. I believe it's going to a Phase 2. They had exciting news release from Phase 1/2. Subjective improvements. They treated 21 patients and majority had subjective improvements in visual acuity. They had poor vision. They are now going into Phase 2 with no results yet. But I believe they are recruiting from 20/80 down to 20/800. And that's not seeing the big count fingers vision. Those patients are still being recruited. Patients with central visual loss, the role there is that the visual cells, when they're injected, we don't expect them to get into the retina and turn beautifully into photoreceptor cells. It might happen. But act as a neighboring cells to increase the environmental milieu, and in fact in the trials they have seen improvement which is exciting. And the other trial, Reneuron. Involve the same cells, slightly different, retinal cells and Jason Commander a good friend of mine, is one of the leading doctors doing this trial. They do this surgery, sub-retinally. Under the retina, inject the congenital cells and they had a very exciting release that they released at RVO this year of some of the data.

Arguably a very small number of patients. I think the number was three patients but all of the patients had, I think, a gain of an average of 23 letters on the EDTRS chart which is a substantial increase of visual acuity at 60 days and 120 days. Sustained visual acuity improvement, even though it is a very small number of patients, they are moving forward as well.

Again, that would potentially target low vision patients so the visual acuity requirements can be looked up on clinicaltrials.gov.

You can see if you're a candidate. But again the diagnosis of inclusion of retinitis pigmentosa which certainly includes patients with Usher. It is not genotype specific.

>> If I can just add to that. Typically in the later stage disease when photoreceptors are already gone, it's hard to make them function again because it's nerve tissue. However, to your point, things like stem cells where you can put new cells underneath or the arduous device or things like that, can maybe help with later stage disease or help functional vision where you can see light.

Scroll to top

>> This question is for Dr. Kenna. You mentioned that for younger children, there is generally no accessibility for the trials. But we have a child who is one and a half two has Usher 2A. What are things we can be doing to establish baselines? Who should we be seeing? What should we do to sort of prepare for the future?

>> Marly Kenna: That's a really great question. In fact I was thinking about the RUSH2A study starts at 8 but we're diagnosing these kids sometimes at 6 months or 12 months. So it would be actually nice to have an extension going downward to younger patients. Some retina data we have in our patients with Usher syndrome, especially 2A, some of them actually have normal retinography for quite a long time. And I think that data will be important to capture on all patients who are diagnosed as very young children.
And then when the treatment and clinical trials begin to show both safety and efficacy, you wouldn't be worried about losing the vision you have. It's quite likely they will start to drop the age a little bit.

As mentioned, usually you start with adults and also because they usually have a more severe form of whatever it is being treated, but I think to be prepared, I think you have to start as soon as they are diagnosed getting baseline data for that particular patient, related to their genotype. Does that answer the question? Yes, Mark?

>> Hello. My question is regarding genetic testing. Would you recommend or is it beneficial for someone who has Ushers to get tested to see if they decide to have children in the future, if that would get passed to the children and what about if they have siblings.

>> That's a very good question. The question is should patients who already have a clinical diagnosis of Usher syndrome, should they or the siblings be tested. There are a couple of reasons for those who have Usher syndrome to be tested. One is to make sure that it's actually what you have. Make sure that you have Usher syndrome. I was talking to patients about correct diagnosis. There are other forms of vision impairment and hearing loss that are not Usher syndrome.

So it is really important to figure that out. And to know that you have the correct diagnosis, especially now that we're talking about treatment trials. If there is a genetic-specific diagnosis, you want to be able to participate in that perhaps.

And the second question is about peoples' other family members. Parents, siblings, probably should be tested if that's available, especially as they become young adults

Scroll to top

because they may want to use that for personal information, family planning or whatever. So I think those are two excellent reasons to get tested.

>> Hi. I'm way in the back. I see a hand in the front. I'll be up in a minute.

>> Hi, so the STELLAR study is targeted on the exon mutation USH2A. Can you talk about what percentage of the exon 13 has USH2A? Because my DNA test just says I have USH2A.

>> Aniz Girach: What percentage of the population have the exon mutation and how we be sure of that and with newer ways of genetic testing, it's much easier to be able to figure out what mutation is there and what mutations are not there, which is another important thing.

Like Dr. Kay pointed out in the natural history disease study, we want to ensure there is population of exon 13 with USH2A patients that we're studying because that reduces the amount of confounding variables in the trial and allows us to be able to better interpret the results of the trial.

So if for example you have any of the newer panels of the genetic testing done, and there are various ways that have been discussed about getting access to those, free of charge often, those will be able to give you and your physician a sense of what mutation is there specifically, and whether you would be potentially eligible for certain trials or not at the primary step.

With regards to the numbers of population, at least conservatively from our records and that is just based on literature searches, we believe there is probably about 16,000 patients or so. I am sure that's an underestimation or so, and you might have some thoughts about this, in your practice, whether you feel that's an underestimation or not. And at least the sense that I get, and at least a few years since I treated patients with Ushers. Talking to colleagues in Europe, and here in the states, it's related to the fact that not everyone has been genetically diagnosed of course.

>> If I can just add to that. I think that adds to the importance of getting tested and identifying the gene sequences, et cetera, that can help with these. I think as treatments get improved and are available, I think more patients will come forward and are tested, and I think that is just natural. Right now in some disease areas, not in inherited retinal disorders, but XUS, patients are automatically sequenced. We don't do that here but I would suggest that it would be good to get that done and as we learn

Scroll to top

more about new treatments moving forward, we can identify that better and have a better idea of population studies that way.

>> Christine Kay: Just to add one quick comment. I'm as you know an investigator for RUSH2A and even I have to go back through my database and remind myself, there is a new exon 13 study going on and let me go through my database to see which patients have USH2A. It does not say in my report exon 13. It's going to be a bunch of gobbly gook. And I have to know that exon 13 is this particular deletion and I have to go back to my patient's genetic report. It's in my database and I have to take the time to do that.

If you have been told by someone that you have USH2A, I would not rely on the fact that someone has done that for you. I would ask that physician and say, can you check? I want to see access to my genetic report for 2299, because it may not say exon 13. You can do a tele counseling visit with InformedDNA and they can explain to you. Number one, get genetic testing and number two, get counseling from a counselor so you can understand your results and what trials you may be a candidate for.

>> Hi. My question is about Usher 3A. And I know more people with 3A than I do with 2A in New York City. So we have quite a few people with 3A and they did do a study, but with a mouse. But I believe the mouse only had a hearing impairment.

And we know our gene and we know the gene that can cure it. The mouse only had a hearing impairment. And I do believe that there were some looking at pharmaceutical treatment. So I want to know what is being done for 3A and any improvement even with advanced vision? If I can just get what I had 10 years ago, with my cochlear implant, my quality of life would greatly improve.

And also, it would prevent any further loss of vision. So that is another thing to consider. Not losing what vision you have.

>> Marly Kenna: Thank you for your question. The question was for Usher 3A. Right now the only form of known Usher. CLARIN1 is the gene. They have been looking at small molecule therapy primarily for the hearing loss but in reading through the fine print of the articles from that lab, they're looking hard for the retinal phenotype because the small molecules seem to be working, to some degree, for the hearing so I think they will go back and look at the retinal phenotype. Hopefully there will be some information to come.

Scroll to top

>> So the question was, are there pharmaceutical treatments being looked at? And I think that is exactly what that is.

>> Marly Kenna: Yeah, so the small -- they're looking at small molecules which are pharmaceutical treatments and looking at libraries of existing already improved chemicals that might actually have a broader applicability of the particular form of Usher syndrome, but right now it's in the laboratory in mice and if it's promising, they will bring them out of the labs and into trials that we have been talking about today.

>> One of you had mentioned that there are other diseases besides Ushers that cause both vision loss and hearing loss. Do you know of any such diseases?

>> Marly Kenna: Actually there are. There is a recently identified gene called TUB4B and the reason I learned about it, I had a patient who thought they had Usher syndrome and looking at the patient clinically, they didn't seem classically like they had Usher syndrome. They have been traveling the country getting testing. And the new gene had been identified in France. And we have a new one. I got an email yesterday about a patient. That's an example, in a patient looking at their genetic testing which had not been positive yet, but in retrospect, new information. CEP290. There are some other genes that are related to vision impairment and hearing loss.

So if you get tested for Usher and the testing is negative or inconclusive or the hearing or the balance or the vision impairment don't seem to match Usher syndrome, it's absolutely imperative to think about getting other testing in. You can target a much wider array of genes to look at X or genomes.

>> Can be an infectious process.

>> Yeah, there can be a lot of other causes.

>> I have a question, I think there is time for one or two more after this. My question is: And I am asking this for a lot of the audience. We have been talking a lot about 2A and treatment coming for some of the diseases. What is it that attracted you guys in the pharmaceutical companies to pick USH2A to be the disease that you're working on and what would it take for other diseases to be what your businesses pursue? I will let you take that first.

>> Susan Schneider: USH2A is of high interest to us, but so are other inherited retinal disorders and other Usher-type syndromes because what we're targeting is a protein function and nonsense mutations associated with that. That's really what is helping us

Scroll to top

target USH2A, in that it has a high sense of nonsense mutation frequency. It makes the most sense to go with those with the highest frequency, and we can go from there. We want to have the best chance forward and that seems to be USH2A for us.

>> Aniz Girach: I will give a more generic reason for this. There are two big reasons that pharmaceutical companies go for this. One is science. And two is the commercial. The science, ultimately whatever chemical or investigative drugs that are being developed, there needs to be some science behind it or being able to target specific mutations in this case or specific diseases. And that needs to make sense from a scientific point of view. That there is a defect there and that defect can be rectified by the particular drug or chemical we're looking at. The science needs to be there first and foremost.

Many of my colleagues in the companies that I work with, also have a commercial hat on. Because typically it takes over $1.2 billion to be able to develop a drug. And so that is typical.

Most drugs actually cost a lot more than that to develop, and typically drugs takes between 10 to 20 years to develop. There is a big investment from the pharmaceutical companies in the long hall and of course the company needs to be able to recoup some of that back into R and D, research and development.

And of course you hear fat cats and big money makers, I don't know if you have seen many, but the world that we live in, is usually very tightly controlled, difficult budgets and you know, we want to be able to ration the money that we have, and channel that money into areas where there is the greatest chance of success. And so as much as, for example, my family members who have specific diseases in the eye, causing them blindness, I have turned around to them and say unfortunately, a company that I am working in, they're looking into a different type of disease causing blindness and these are the harsh realities that we have to go through as physicians and working in these companies.

But science, first and foremost, it should make sense. That's the greatest chance of success and ultimately there is a way in which the companies need to make the money to plow back into further research and development, and I think that's really important to understand.

>> Thank you. We have time for two more questions and I see about ten hands. I'm going to try to do my best to get a couple of you here.

Scroll to top

>> Hello. My question goes back to the genetic testing. I was testing about 12 years ago in the State of Texas where I live. I have USH2A but I now have a great nephew, so my niece's son who is in Georgia.

My question is, how important is it for him to be tested by the same company I was tested with? Does that make a difference?

And also, do you all use any of the databases from these companies that do the genetic testing to help recruit for your trial and your study?

>> Christine Kay: I'll start. So the question was regarding genetic testing. She has a great -- what is it again? Great nephew who lives out of state, who I am assuming is young. 10. And I am assuming he is unaffected. And she wants to know -- so she has Usher. The lady who asked the question has Usher and wants to know if the unaffected 10-year-old family member should be tested. This is a very interesting question. And I will try to be short because I know we have to go to lunch. There is a caveat of genetic testing in asymptomatic minors. I trained with Ed Stone at the University of Iowa and he has a long talk about whether to test asymptomatic minors. There is a lot of controversy on that.

I think if there is potential treatment trial like Luxturna and we have a potential risk of them being affected from a family member, then yes but if there is no current treatment or technology that would change any management, there is something to think about with testing asymptomatic minors who are again, not symptomatic.

It's important for that child to see an eye doctor, a hearing -- you know, a pediatrician, and follow them like a normal patient. If they have any evidence of any issue, vision wise or hearing, then absolutely genetic testing. But if it is USH2A, we know there are some treatment trials going on right now, then the question comes down to, how likely is it that that family member is actually affected?

So this is an autosomal recessive disease. For him to be affected down the family tree from you, your risk of giving it to your offspring is relatively low and the pedigree gets lower as you go down. And there is a decent amount of USH2A in the population that were carriers. It is quite confusing. However, regarding the cost, you wouldn't do a whole panel on him. If you wanted to and his family wanted to rule it out, you would just test the USH2A gene. It does not have to be done at the same lab you had yours done at.

Scroll to top

Labs share information. You have your information as back-up. And you are the pro-band. The affected family member. Most insurances will not cover that. It's hard to get that grant funded because most insurance companies require you to be affected to cover the cost. It can be hundreds of dollars, up to $500, I am just guessing, to just test the USH2A gene commercially out of pocket, knowing that no insurance company will cover it. We can talk more about it at lunch. You can get the InformedDNA, the tele-counseling service involved. That's their job to give you an answer to that question. Great question. And that's what they do for a living. I would suggest genetic counselors. And it's incredibly different for the ages and the patients that we're talking about.

>> Aniz Girach: To chime into your question about whether pharmaceutical companies [have access to the genetic testing results databases]. We don't generally because of HIPAA. As pharmaceutical companies, we rely on physicians like Dr. Kay, for patients in their databases. And also in group meetings like these. We were talking, Krista and I earlier, our Phase 1/2 trial, STELLAR trial is actively recruiting patients now.

It's a crying shame that we have a house here of patients who are potentially eligible and there is not an easy route for you to get into those trials. I would urge you to work with the Usher Coalition because that's an easy way to get connected yourself and to the investigators that are working in many of the trials ultimately. Really a strong plug for you to get involved in the Coalition.

>> Christine Kay: Final point on that. Do they sell your information? I'm aware of two comprehensive panels for free. Foundation Fighting Blindness and 366 gene panel through InformedDNA genetics and you sign up through My Retina Tracker with the foundation. Foundation Fighting Blindness is not allowed to sell the information. Carefully written in the IRB. But you can call them, how many patients do you have with USH2A. They can tell you a number, not give your phone number. ProQR can say, can you please distribute my flyer that I am doing on USH2A to the patients in the my retina tracker. The patients will get a flyer in the mail that a company is doing a trial. It's great. But they are not selling your information and another is spark Therapeutics. 250 gene panel. You get Invitae by a genetic lab. This opened up two or three weeks ago. You have to read very carefully, the consent, because they do potentially sell your information to pharmaceutical companies or other companies. I'm not aware of where it goes but that's much more of a commercial pharmaceutical access to information. It's up to you. But those are the two ways to get genetic testing for free with counseling.

Scroll to top

>> Mark Dunning: Okay, I know there are a bunch of other questions, including you Andrea. But we are late for lunch by a lot. What we're going to do is take a break now for lunch and get together at 2:00. Lunch is outside. Outside the back. Go out the back. So go out the left or go out the back. Okay, so go out these doors and take a right and around and there is room over there to eat. We will see everybody at 2:00.

If you have more questions, please, guys, they are around. Thank you very much, everyone.

[Lunch]

Scroll to top

"Importance of Collaboration in the Educational System: Identifying Your Team Players" Lanya McKittrick, PhD and Carly Fredericks

>> >> Hi, everyone we're going to get started in just a couple of minutes. If we can ask everyone to take their seats. All right. If we can get everybody to take your seats. We're going to get started.

All right. So if everyone can take their seats, and I will ask the interpreters, if you guys are ready to go? Okay.

Thank you, everybody. Hope you enjoyed lunch, and we're going to get started with the second half of our program, which is going to focus more on the family side, and less on the science side of things.

Our first speakers are going to be Lanya McKittrick Ph.D. And Carly Fredricks, who are both parents of kids with Usher syndrome and going to talk about kids with Usher syndrome.

>> Thank you so much. How is everybody doing? Okay. Great. I'm just going to do a quick accessibility check and make sure that everybody can hear me, captioning is working.

So we are plugging our Roger pen back in, Lanya and I are stuck together. She is going to get sick of me. Welcome, everyone. On behalf of Lanya and I, we want to thank you for the opportunity to share some information with you today.

I'm starting to notice a pattern. Every conference that I'm asked to speak at, they ask me to present after lunch. I don't know if it's because I have that Jersey girl speech, to entertain you and help you not fall asleep, or help entertain you until happy hour.

Welcome to the conference. If your child has recently been diagnosed, we want to welcome you with open arms. We know this can be an extremely overwhelming time for you and your families. Lanya and I love to share our stories. We know it's very different and we find comfort in knowing that we're not alone. The Coalition provides us the opportunity to connect and it's so important to us.

I'm going to start off, you can see on the screen top left, or your right, this is our family. My daughter, her name is Ava. You may be familiar with the organization Ava's Voice. We run a non-profit in the State of New Jersey and now we're national. And Ava has two families. So we do co-parenting which is a difficult thing in itself but we found a real balance to be able to put Ava first and work together, not only through the diagnosis of Usher syndrome, but just raising a child in two families.

Scroll to top

So we are very fortunate. That's Ava's dad there. James, his wife Amber, Felicity, and another baby sister coming in December. She is excited about that.

There is myself, along with Ava's step father, Paul who is here with me today and then Mason and Miley.

>> My name is Lanya. I am honored to be here with you. My family is also up there. Family of four boys. Conner, Coal, Hunter and Dalton. I have two boys with Usher syndrome. Conner is 20 at the Rochester Institute of Technology School For the Deaf and Dolton is 11 and going into the sixth grade.

As Mark said, I just got my Ph.D. Thank you for the plug. As a parent, I have been trying to figure out this journey on what works best for my kids working with the schools and we had a lot of struggles so I went back to school to get my Ph.D. in special education, focusing on deaf blindness. What we will be doing today, Carly and I will be sharing a little about our stories, experiences, as well as strategies that worked for our families, and wanted to present a little bit of research that I have done in the field of working with IEP teams, educational teams. Working with kids who are deaf-blind.

So the presentation objectives today, we're going to present parent-initiated strategies advocating for children while working with IEP teams and understand the challenges that children with Usher syndrome experience in the classroom because of the dual sensory loss.

So a little bit about the study that I have done. Primarily like I have mentioned, my research has been really focused on working with families. And one of my primary goals is to not only help families, but also bring this research back out to the educators and professionals that are working with our kids.

So that's why I feel it's a really important topic. This year for the last couple of years, I have done several research studies in this area, and I am honored to be presenting it not only to you guys today, but also to a lot of educators in the field of deaf blindness and special education overall. And my goal is to increase awareness of the need for more professionals in the area of deaf blindness, to hopefully increase the outcomes for our students.

So the study that I'm talking about today, the purpose was to really look at, I mean we know, Carly and I have talked a lot about the strategies we use and the challenges that we have had. I wanted to talk with other families to see if they have the same challenges. So the purpose of the study was to look at the strategies that parents of children who are deaf-blind, what do they use when they foster those collaborative relationships with the IEP teams?

Scroll to top

Part of the study I interviewed is mothers of children who are deaf-blind and I interviewed them in late 2018, early 2019.

The research questions I had were in three areas. First off, what experiences lead parents to develop and use collaborative skills to work with the teams? And what does it mean working with the parents and the teams? And lastly, what strategies used to promote collaboration? What can parents do to improve to make the meetings more collaborative when working with the teams?

So what I found was not surprising, because it is what my experience has been as well. It's that vital role that parents play in the meetings. Case managers. Advocates. They share knowledge about deaf-blindness because of the disability, and when I did my research -- I'll be the clicker. Sorry.

>> We're navigating. There we go.

>> Thank you.

>> There are three categories that emerged. The problems that parents experience. The collaboration and the advocacy strategies. So a little bit about the problems that parents experience. So probably not surprising to all of you in this room because you live it every day, is that there is a lack of knowledge about deaf-blindness and the unique student needs in the classroom so what you have seen if you look at the research, is that there is a lack of team expertise. I know from myself with my kids, we don't have a professional who knows anything about deaf-blindness on the team.

We have a teacher of the deaf, teacher of the visually impaired, but really I, myself, am the expert on deaf-blindness on the team and there is a shortage of qualified personnel that are experienced in this area. Also not understanding and valuing different communication modes, and that may change over time. It may depend on different situations. Tactile sign language might be used. Sometimes may not be necessary. Other strategies. And really understanding and valuing the different communication modes and the heterogeneousness of deaf-blindness.

A lot of times I talk to families who say, well, there was someone in our district who understands deaf-blindness but they don't understand a child like mine. They don't understand that my child is losing their vision, but their vision is actually declining over time so what might have worked a month ago, might not work now.

And there is a large team of service providers we're all working with. That can be a challenge because you go into an IEP meeting and there are so many people to collaborate with.

Scroll to top

Lastly, difficult transition. Not only transitions to post secondary, but transitions every year to new team, new teacher, things like that.

And also district-level issues such as lack of resources, compliance and implementation issues.

In regards to collaboration, we know collaboration is vital for supporting our children and the knowledge sharing is also equally as important. We will talk later about strategies and knowledge sharing that we bring to the table and the teams. Because we live it every day and working with our kids, it's important to be in an environment that is collaborative so we can share that knowledge with others.

So this is what we're going to get into a little bit. Advocacy strategies that we bring to the table as parents, there is direct strategies such as, you know, things like pre-planning, asking for information in advance at the meetings so there are no surprises, advancing the meeting so it stays on track, appropriate goals and ensuring that our child's needs are met.

And also what I call leadership skills, remaining positive in the meeting. Building consensus and having this team mindset, thinking ahead. Being firm about what your child needs but also being flexible, listening to others and really playing that leadership role. And lastly student involvement.

I know for myself, I have a 20-year-old. His transition planning did not go all that well so what I learned throughout the process is, how can I involve him earlier on in the educational decisions so he can become an independent adult who is making those decisions for himself? So I worked really hard as a parent and found the benefit of involving him early on. At the age of 8, he would be going to meetings himself, talking about how the school year is going and we have seen a lot of benefits in him doing that.

I'm going to turn it over to Carly where we talk a little bit about -- how does this translate to practice?

>> There is a reason I opted out to run the USH summer mini youth camp, because the research goes way out of my ahead. I thought, I will take 30 kids out in Philadelphia and entertain them while you guys do research. If you guys are ready for a nap, I am definitely ready for happy hour today. So we are going to bring, you know, it was really important for us, okay, it's great. And research is wonderful, but who is going to support me day to day?

I would find myself coming to local conferences, going to daily trainings and feeling motivated and so excited. I knew I wasn't alone and they taught me some of these

Scroll to top

things that I would share with my team. And I would go back to the team and they may not be receptive and not understand Usher syndrome as a disability. I sometimes call it an invisible disability.

They may be playing on the soccer field or in high honors classes but they can't understand why they can't see at night or can't hear their friend calling their name from the other side of the reason. I always say the disability isn't the problem. The accessibility is the problem.

I have had case managers saying, she doesn't need an IEP. She is functioning on grade level. She is actually on grade level. But without all of the support and specific accommodations in her IEP, she could not be as successful and it's so hard and almost defeating for parents to sit there and try to convince the people across the table that it's such a need.

We really want to talk about that today. Taking the data and bring it to practice. I'm going to tell you a little bit about Ava and thanks to Usher syndrome Society for capturing who Ava is. That's Ava right there. For those who can't see the screen, she is wearing a blue shirt and gray shoes on and legs crossed and she is confident. Don't tell her she can't do anything. Because she can and she will.

I wanted to show you an overview of what it looks like in the life of Ava in the past 13 years working within the educational system. From birth to three, we received early intervention services.

At that time we did not know that Ava had Usher syndrome. She had failed the newborn hearing screening. I was given a pamphlet from the hospital that said, how do you raise a deaf teenager. I said when is the next time you need to eat, when do I need to change your diaper. I was worried about raising an infant, not a teenager. When I found out she was deaf, I wanted to find out options and we worked with a school for the deaf and also someone from a speech school who had services as well.

Privately we had to attend a local ASL program that was actually for hearing children in a very wealthy neighborhood that thought it was a luxury to teach their children sign language. And that was my only option for being able to teach Ava sign language. That was a little overwhelming for me. Because there I was really trying to connect. I was trying to learn. And these moms really were just trying to have something fun in their day that they could say was a luxury for their child.

I found myself feeling a little bit alone and defeated at that moment. And I was really on a mission to advocate for more, because that more that we needed at that time was not available.

Scroll to top

After Ava aged out of early intervention, she then transitioned to the Summit Speech School, auditory oral program at the time. She received teacher of the deaf services, speech services, physical therapy and that was the time that Ava was then diagnosed with Usher syndrome.

I had gone to her 30-day review meeting and we sat down. Everybody was all excited. They said, how is it going? Well, the hour-long car ride is a little overwhelming for me, putting my baby on a bus. But I knew it was the best placement for her. They said what's going on? I just said I don't know, I feel a little funny. I feel like she may not see me at night. They said you know what, have you ever heard at Usher syndrome? And the director I thought was going to fall off her chair, because the teacher had mentioned that word.

And she said no, no, no. I don't want to tell you anything today. We're not diagnosing. But the minute I read it, that's when we knew Ava did have Usher syndrome. We did confirm it and thought at the time we wanted to expose her to more resources. From 5 to 10, main streamed in our local school district, received the itinerant services and started working on orientation and mobility, other ways to do private PT.

She said hey mom, I'm never walking on a balance beam. I hate to tell you that. I never with will. I don't want to do PT anymore in the school. I said okay, let's get creative. You have to do something. We thought about other ways and did HIPAA therapy with horses. And really worked on her core training and age 7, transitioned to a new school district. Overwhelming time to educate peers and professionals. We said to the district, we want a treatment plan. We want to not only start working with the teachers that we will be working with now, but throughout middle school.

I had access to the professional that is we're talking about, that had the deaf-blind expertise so collaboratively with the teacher of the deaf, TBI, deaf-blind specialist and myself, as well as Ava, we trained the entire staff for the next three years and today, Ava is going to start preparing for transition at the age of 14 working with a VR counselor.

>> So just a little bit, because I have two kids, I'm going to do a really brief overview. Our experience is pretty similar to what Carly described for Ava. What I'm going to say is -- I'm not going to read all of these slides by any means. The slides are available.

What we did for our kids -- Conner started early intervention in a toddler program, went to preschool. Like Carly was saying. And then we found out about the vision loss at age 5. It was very similar.

At that point he entered elementary school and was in a private school setting. At some point a few years after that, you know, we were handling everything ourselves.

Scroll to top

We didn't really have an IEP for him, an educational plan for him, and we moved him back to the public school. What I would say is, we sort of looked at what is the need for each year and we weren't afraid to be flexible if we needed to. We moved in and out of private schools, back to public schools. Moved public schools several times. Did it thoughtfully and in collaboration with the educational teams that were working with our kids.

But you know, some years were good years and some years we really struggled and I think it was because the vision loss was changing over time, particularly in the late elementary years when we really started to see some peripheral loss, when we started to add all of these services, Braille, orientation mobility services and had all of these things. How do you keep him in the classroom as well as provide the services? That was sometimes challenging in a private school setting because there was not enough time in the day to do everything.

One of the best things we did was just remain flexible. We weren't afraid to really change the placement in the school and the environment that our kids were in, and that really was a benefit. As you can imagine, that's really hard on a kid to always be changing.

Hard on a social perspective. Hard where you have new educational teams that you're always working with, and really just building those relationships with those teams was really vital to the success that we had. Same thing happened in middle school where it was mostly public. But again, no point person, so a lot of the times that I was bringing all of those team members together, and trying to problem solve, figuring out what the best strategies were for that particular year. And then high school, he did move back to private school. It was a really good environment for him. It was a smaller setting and it worked for him, and he had a really supportive private school team and I don't think it really matters whether it's public or private. It ended up where we had a really supportive team and did not know a lot about deaf blindness and didn't know about Usher syndrome and willing to learn about it and willing to listen to Conner and our family about what the needs were.

Dalton had a lot of experience. Similar. A lot to coordinate. We recently moved him out of private school into a public school setting and it was a really positive experience. We really focused hard this time on self-advocacy, self-determination and making sure that he is equipped, like mentioned earlier, with the skills necessary to make decisions for himself on what is working. That's something we really tried hard to do as a family, to listen to, even at the age of 11, what is working for him in the classroom and made a decision to move schools to a different middle school this year because of what he wanted to do and what he was telling us.

Scroll to top

>> So breaking down those barriers, I think we can all agree that's the hardest thing. We are working right alongside of our children and the teams as well. Sometimes you don't know all of the resources available. One of the things that I have found to be so instrumental in Ava's success, people say, how do you get the team to agree to certain services and how do you get them to agree on bringing in outside itinerant programs? To me it's about identifying expectations. Sometimes we walk in and everybody's walls are up. District may be feeling the pressure of not having financial resources, have a budget. Not sure what you're going to ask. They know you can be a bull dog in the community and an advocate. They're nervous and we're nervous. Such an important decision and to me, in this moment, I can't fathom that we always get in a room and advocate half hour or an hour in a meeting to decide the next whole year of your child's life. It's so unbelievable to me and then we are so afraid to sign something because what if we want to go back and ask somebody something. All I see is your mouth moving and I am so overwhelmed right now.

I try to break down the barriers. I say listen, we all have a level of expertise that we're bringing to the table. I need you just as much as you need me. I am here to share with you everything I have learned about my child so we can make a decision together and sometimes that immediately puts everybody at ease. We don't expect everybody to know all of the answers. How many of us are running to different audiologists, ophthalmologists and now there are vestibular issues. We're all over the place. There is not one person that brings the dots together.

That's our role as parents and setting up expectations is really important. And that goes right into setting the tone. They want to feel needed, too. They worked really hard to get to their position and at times that I may not agree, I would say, you know what, let's look at that again or let's table that for a minute so we can all sleep on it and maybe come back and have a future conversation.

Again, sometimes it's setting that tone. I'm here to work with you. I'm not here to disagree or say it's one way or no way. Let's set the tone and say we're going to continue to work through this, even though we may not agree at this time. For me personally, I'm always using challenges as teachable moments. I actually say Ava is transitioning into eighth grade so we, every year, decide to meet with a team that we know will be next year, with the understanding teachers change, teachers aren't available, classes overflow happens, that's okay but we want to try to get all teachers that she has potential to work with next year, and have a meeting. Say hello. Put a face to the name. We know them getting that document, they don't know who our kids are. I'm sorry, I hate reading through an 80-page document and I don't blame them if they don't want to read through it. It's a legal document. But I would want them to implement them. I put everything on the table. I want you to know that next year is a

Scroll to top

new year, and Ava's vision is changing and there may be times I update the team. If you reach out and me and I'm not sure what happens, I may CC somebody that may give you an additional resource.

I don't have all of the answers as the parent but I don't want them to feel that I am CCing everyone and their mother to get them in trouble. Using the challenges, sharing with the team, and say please don't be offended if I CC somebody. But if we caught something here that can help a future teacher in another classroom, we want to make sure that everyone can be included.

This is my favorite topic, right, Lanya. You don't know what you don't know. People say what vision doctor do you like to use? You know, I like to bounce around. Some say I'm crazy. Our state, Commission For The Blind, every two years Ava can get a new low vision examination. I like to visit new doctors because there are nutritional ophthalmologists, those that create new lenses and those that use therapies that they think helps. I like to learn something new. I like learning.

Sometimes our kids are so, so good at faking it until they are making it. Ava will say, I'm fine. I'm doing great in the classroom. I don't need my FM system. Tell me why not. I can hear that person clearly. Then we start to notice that she is missing things and her work starts to decline a little bit. I don't know, I thought that's what he said.

And our kid, it happens to them a lot, they don't know what they're not hearing and don't know what they are not seeing. A lot of times we're working alongside our kids to kindly remind them about the resources and the importance of utilizing the resources that they have available to them.

Because for them, for me especially, Ava doesn't come to me until she is feeling totally defeated. She is exhausted. She is trying to listen all day. She is trying to focus and she is overwhelmed and it isn't until that moment where she worked herself so hard straining by not using her resources, that she just then falls apart.

>> I would say, I wanted to add, I think communication is really key to really ask what is going on with your child. I had the same experience with my kids. They come home and everything is fine. I'm fine, fine, fine. Keep drilling at it and listen to what they say.

Sometimes I have that mom instinct that something is just not right or something has changed from where it was two months ago. We don't know. Maybe there is a glare in the classroom. Lots of times they move their desks. Like every month they move to a different location. Maybe that location is staring at a glare and aren't able to see something. I am always trying to ask questions of my kids, hey, what's going on? Did you move desks?

Scroll to top

Are you sitting by somebody new? How is it going? Communicating with the teacher and communicating with the entire team constantly. Like Carly was saying, you're meeting once a year to figure out that year but once a year is not enough. Particularly when the vision loss and hearing loss is changing over time and the settings are changing, you have new people coming onto the team and it's the constant communication that is really important.

>> Same for us as parents. Asking questions. What are my options? What type of programs do you have available? A lot of times things are presented to us and we think, oh, that's what they have. That's all that is offered and don't find out until we start talking to our neighbors, at the soccer field and talking to another mom and say wait, I didn't know they had that program.

Just keep asking questions. What are our options? I had something envisioned in my mind that I thought would be great for my child but I don't know what exists in our town. That helps to make more educated decisions as the parents. And then for the educated professionals, they feel like they are struggling. They don't know the deaf-blind specific resources and don't know all of the service providers so we find that as the parent, it's our role, if we are knowledgeable with a resource, to share that with a team. I always put it in a way, hey, I have this resource available to you. Let me introduce you. So that way, they can take the lead for the next step for more opportunities and training. And again, understanding the unique needs for a child with Usher syndrome.

With Ava, now that she is a little older and we're meeting with the teams at the end of the year, she is talking about herself and sharing about herself.

This is Ava in a nutshell. I like to show this at every presentation I do. It's a recap. I like to show a lot of information. It's a lot of documents. What information do I need to know? They have another 20 to 30 kids in the classroom that also need support. The portfolios I do, it's the "A's" of Ava. She is athletic, artistic, advocate. I want them to know who she is as a person first, and then I really like to pull all of the specific things that help her on a daily basis, to recap Ava in a nutshell. Instead of going through this whole document on your own, I will tell you what she needs.

She needs her FM. Needs captioning for videos in the classroom. May need to have peers repeated. May need extra time. May need to leave early to get to the next class before the herd of students. The teachers appreciate it and they say it makes it so easy to look back at the paper in the desk. She hasn't been using that FM. Let me see why she isn't using it.

Introducing teachers to their own colleagues, they are their own support system. They have access to the teachers daily. They can walk down the hall and say hey, you

Scroll to top

worked with Ava last year. She is having a difficult time. She doesn't seem to be able to read the maps in social studies. What did you do? Don't want people to reinvent the wheel. Just do what is working.

>> I know the professionals working with my kids are itinerants and they're not actually located in the school themselves. They are coming in on a weekly basis or whatever basis to work with my kids. A lot of times working on self-advocacy skills and other skills that they are working on. I spend a lot of time also just with the transition plan, making sure that everybody knows what each other is doing so we're making the best use of the child's time as well in the classroom and outside of the classroom so they're not missing information by not being collaborative.

>> And then we say ride the wave. You know, a lot of parents ask me, when do you talk to your child about the diagnosis? Or when are you reaching out to the teacher of the visually impaired throughout the journey of Usher syndrome? There is no single plan. Every single person in this room is on their own journey. For us it's to share the resources with you and you decide what's best for your family and your child.

Sometimes you will find that for Ava, the teacher of the deaf was the go-to person I would say from the time she was little up until about fourth grade and then TBI stood up to the plate and she was that contact. Her vision has been impacted and hearing on the back burner. We figured out what works. She is thriving and working on the vision.

I have been bringing Ava into the meetings since second grade. I told Lanya a story the other day, the first time Ava came into the IEP meeting, brought her in for the last ten minutes, what do you think about second grade? Anything you want to change for next year? She said yeah, I have a little bit of a problem. Maybe we can change it. It's not a big deal. We thought oh, my gosh, what's going on. We didn't expect her to talk.

And when we do the lock down thing, I am put in the back of the room, but my cochlear implant gets stuck to the back of the file cabinet. Oh, my gosh, has this been going on for six months. We wouldn't have known. Otherwise, would have just dealt with it and think no big deal. I thought, you could have just told your teacher and she could have moved your spot.

A lot of times, they don't want to inconvenience anyone and now I am stepping back and letting Ava take a lead. It's important for both of our kids.

>> We have had some challenges with our teams. And I find that it has been helpful. In times when I really struggled with getting them to understand, we have pulled back

Scroll to top

and I put my kids and let them describe the situation and it's a lot easier for them to describe it on their own.

>> So the last, what we're going to leave you with, I satisfied Lanya, I don't know how we're going to do this presentation in a half hour because I can do this in three days. Quick and dirty version that we will leave you with. Bearing good fruit. There are times I have asked teachers, if this isn't good for me, just tell me. I am not here to get you in trouble. We can figure out who can handle this. Educate and involve your child early. It's really important. Have those conversations. Being on the offense, not the defense.

To me, that planning ahead is so important. If you're ready for the following year, you are not feeling like you're playing catch up and overwhelmed. Don't feel like you have to be so creative. If there is something available that we can adapt for your child, we're happy to do that.

I was hoping to have like a mic drop moment, Lanya, we talked about this. Wanted to have balloons up here that say teacher of the deaf, interpreter, lunch aid, bus driver, all of the people that you're talking to on a daily basis. You are the balloon together and hoping to let them fly. I don't know if anybody knows. I'm living under a rock. There is a helium shortage in the nation. I had no idea. I was living under a rock the whole time.

You're the balloon collector for your children. It's your job to pull the resources together and get them talking together. It's a team. Without the team, our children are feeling isolated. It goes back to the first thing we said. Accessibility. Communication is everything. That's what we're advocating for and that needs to happen from the beginning.

>> Yeah so we're leaving you with a list of resources and like Carly said we can talk about this for days. Both of us love helping families. I just scratched the surface of my research, but if anybody wants to email me, I am happy to share details of this study or previous studies I have worked on with families who are deaf blind. Feel free to send me an email. My contact information is on there. Carly feels the same. Reach out to her as well.

>> Quick plug for Patty McGowan, Nancy O'Donnell, a few years ago, we did a study on Usher syndrome and there is a link, and it dives into intensified strategies about services that our children were receiving. Feel free to connect with us. Go to the tables. We can share the direct links. If you have questions, please, please, please reach out to us. We do run a family to family call with NCDB and if you can join us, I believe the sign ups are this month and next month and then we can connect with you

Scroll to top

on a monthly basis to talk openly and have an open dialogue. Thank you for your time. Again, welcome to the family and we look forward to meeting all of you in person.

[APPLAUSE].

>> Thanks, Lanya and Carly. They were here to talk about the needs of kids with Usher syndrome and ways we can help them. One of the big challenges that adults with Usher syndrome face is employment. Particularly there is a very high rate of unemployment among people with Usher syndrome and our next speaker, Ryan Thomason is going to talk about the benefits of employment with the federal government for people with Usher syndrome and we're going to take just a second to get him set up here and then he will talk to you.

Scroll to top

“The Benefits of Federal Employment for People with Usher syndrome" Ryan Thomason

>> All right. Hello, everyone. My name is Ryan Thomason and I am going to talk about federal employment for people with Usher syndrome and hopefully you guys can get something out of this.

The legal disclaimer, that I had to say. All information in this presentation is publicly-available knowledge and I tried to give you all of the places where you should be able to find that within each slide.

And I am not doing this in any official capacity as an employee of the federal government.

So quick about me. After college, I had 10 years of employment in the private sector. I have spent the last three years as an employee of the Department of Defense, here in Philadelphia. I have been married for 14 years. My wife Jennifer, we met in college. I have two kids, Lincoln who is 11 and Finley who just turned 9.

I was diagnosed With Ushers 2A when I was 28 years old. I had always loved sports. I grew up playing sports. I was a three-year varsity starter in football. I did track and field for all four years of high school and I ended up doing it for three more years in college as well. And yes, I am a nerd. So it's just easy for me to get that out there.

Making sure that I am advancing the slides at the right page. So I don't know where it's at. All right, what this presentation will cover. I would like you to know what the Americans with Disabilities Act is and how it applies with the hiring process for the federal government and how it filters down in the state and local governments and private employers.

Schedule A Hiring Authority and USAjobs.gov, the main hiring website for the federal government. How the workforce recruitment program benefits college students and recent graduates with disabilities.

And then I will finish talking about the Ability One, what that program is, and how nonprofits can use it to employ specifically the blind. So the Americans with Disabilities Act or the ADA, it was created in 1990. It is a civil rights legislation, kind of molded after the Civil Rights Act in the 1960s, basically giving people with disabilities guarantees from discrimination.

And making sure that we have the exact same opportunities as anybody else when it comes to hiring, where you work, where you go, where you buy stuff, and also, you know, going into facilities and everything like that. So there are three main titles in the

Scroll to top

Americans with Disabilities Act. Title 1 is the employment. That deals with private employers, federal government, state government, local government, trade unions, and it basically gives us the rights that we cannot be discriminated against when it comes to hiring, the hiring process, or anything within that avenue.

As long as that private business has 15 or more employees, it does apply to that business when it comes to hiring. Title 2 applies to the state and local governments. This is basically what your rights are going to the state when requesting services or requesting access, and even down to your local city level, it all applies for the same thing.

Title 3, probably the thing that you guys would most experience, the public accommodations and commercial facilities. Any private business that is open to the public, Title 3 applies to them, when it comes to disability and what people are allowed to do.

Where people take a guide dog into a restaurant and the restaurant says, you need to take your dog out of here Title 3 of the Americans with Disabilities Act, gives you that right to go into that business establishment and eat there. So they cannot discriminate against you.

So if you feel that you have a complaint for any one of these three titles, right on the main ADA.gov website, there is a big huge button for "file a claim" and it also shows a list of all of the current claims that are happening.

So if you ever feel like you don't have a way of fighting back against what's happened to you, ADA.gov website has a lot of that available for you.

So the Schedule A hiring Authority, this, for the federal government is the main documentation and the main process that people use to become employed within the federal government. This is how I became an employment of the federal government three years ago. It is an exemption to the traditional hiring process where you put in your resumes and you get stuck in the pile and hope that they pull your resume off the pile and look at it.

The Schedule A Hiring Authority basically shoves that aside. People who are Schedule A have top priority. If the supervisor looks at it and says this person can complete this job, then you will get the position.

And how it worked for me, I received an email, and they said, would you like to come move all the way across the country for this position? Can you perform these duties? I gave her a phone call 10 minutes later. I told her what I have been doing in my master's program and how it applied to the position. Out of the 10-minute

Scroll to top

conversation, that was four minutes of it. And the other six minutes, we talked about her vacation. She was going to New Orleans and she was going to drink hurricanes.

Because she did not ask me interview-type questions, an hour later I did have a firm offer. We had to move totally across the country. Has been a trip for our family. But the Schedule A Hiring process, does give people with disabilities a leg up.

What you need for a federal job opening. Obviously you want to have your resume. The government has resumes in a different style from what everybody teaches you in school, the one page with very specific bullet points. You don't use any of that. The government uses a long-form resume. If you have two or three paragraphs, basically you describe out all of your job functions that you performed and how you performed it. That is how the government reviews all their resumes. My current resume is four pages long. They tell you not to do that in college. But when it comes to hiring in a federal position, that is what you do. And you have got to make sure that you have all of the documentation required for the vacancy. You find this on the USAjobs website. If you do not have all of the documentation, they will automatically reject your resume. Make sure you have all of that. And the most important thing is to make sure to have the Schedule A documentation. It is what they call a Schedule A letter. Basically your proof and unless they noted that you have a severe intellectual disability, severe physical disability, psychiatric disability, for us with Usher syndrome, we have the vision and the hearing part.

How do you request this documentation? You can get it from your doctor. Licensed medical professional. If you are using a stable vocational rehabilitation program, that's how I got my letter, they can write the letter for you. Each federal state agency that can provide disability benefits, it doesn't have to be a complete break down of every single aspect of your disability, it's basically a professional saying, this person does have some limitations, but they are fully, there is nothing wrong obviously. They are, you know, good for federal employees.

And you can also find all that information on the USAjobs.gov website or a different location which I will show you guys later in the presentation.

So you have two approaches when trying to use Schedule A Hiring Authority. First go to USAjobs.gov. Almost literally every single federal position and every single state and country in the world is on USAjobs.gov. So what you want to do is take all the -- like I said before -- your resume, documentation, Schedule A letter, click on the vacancy and then go through the process of uploading it through that.

Or you can go directly to that federal agency's website. Go to the Department of Labor, and they will have what the career openings are, or the Department of Agriculture, whatever it is that you have a career in, Army Corps of Engineers if you're

Scroll to top

an engineer. You can go to the website directly and it will show you where everything is in the whole country.

Make sure you apply for the positions as soon as you see them. They are typically only open for one to two weeks. They don't leave them open very long. You try to get the stuff as quickly as possible so when you do find an opening, get it in there as quickly as possible.

The second approach is applying with the agency using the Schedule A process. Most agencies will have a disability program manager or a selective placement program coordinator. And these peoples' role is basically to look for people to have disabilities, take stuff from federal databases.

If you have put your name into them already, and to try to accommodate anyone with disabilities for the hiring process. If you go to the link on this slide, that I have included on it, it will show you every single disability program manager at every single agency in the entire United States.

So wherever you live, there should be at least someone, in a lot of cases, DC or Virginia, kind of in this general area where there is a lot of federal employees, you can find a lot of those coordinators.

So the workforce recruitment program or the shortened of it because the government loves acronyms is WRP so you go to WRP.gov. If you are a current college student or you're a recent graduate, wherever you are in your college career, even if you're still getting your bachelor's, master's or Ph.D, as long as you're in college, you can join the program. It's disability employment policy group and the Department of Defense also coordinates with it as well. Defense contractors use this program to find people that they are looking for internships or if they are looking for someone to hire, they can use this program to hire individuals.

So students, they need to go to the WRP.gov website. And with the help of your disability counselor or student counselor at your University, they can help you get set up with an account. And through that, after you sign up, usually late August right before school starts for college students, and a person that is a federal employee will serve as your recruiter. So if the fall, what happens is we put in our application as a federal employee to volunteer for this.

We work with the colleges and given up to 20 students to recruit or to interview and through the interview process, I will basically be asking the student just tell me about yourself. What is it that you are looking for in a position within the federal government, are there any accommodation needs that you're going to need.

Scroll to top

We're not going to ask about your disability or anything like that. Don't worry about that, about feeling uncomfortable. If you don't want to discuss what your accommodations might be, you don't have to.

Basically what we do as a recruiter, we take you to the interview and base all of the answers on a points scale and then we put that and we write a nice little summary and if that person is good enough or if we feel that person is eligible, we will put them into the WRP database and that database is the main database that the federal government and contractors use when they are looking for students that are looking for an internship or looking for a position after college, they can go for one year and go in there, do their search by the state, and it doesn't show anything about the disability. It shows that the person has one and there are no specifics on it. And they can either interview that person or, you know, directly place them into a position.

The Ability One program, this is actually one of the biggest programs for people that are blind specifically in coordination with National Industries for the Blind and Source America are big players in the program.

What this is, the federal government has an obligation that whenever a service or supply that they are contracting out, if Ability One, National Industries for the Blind or Source America provide the services or supply what the federal government is looking for, we are obligated to take a look at them first and accept that contract under them.

If we don't use them. If it's something where the scope is a lot bigger and it doesn't fit within the program, we will pick a different company or something.

But these programs do get the top priority when it comes to getting a contract from the federal government. The National Industries for the Blind is the largest employer for the blind so if you can look around, there are non-profits that basically feed into this program. And that is basically what they do for employment. You put together pieces and tools or if you're an accountant, or you do something in the contracting field of writing contracts, law or anything like that, if there is a service that you can provide and you are blind and can try to work for one of the nonprofits and organizations, they do get contracts from the federal government.

And it is something that at least I found in my field, as a Department of Defense employee, that it's something that, for a lot of the stuff, these programs get picked first when it comes to the contracts.

So Ability One, like I said, use the purchasing power of the federal government, and these programs are dedicated to training and employing individuals who are blind. If you're looking for different kind of skills, these programs will train you for that. And then after you have received training, they are trying to help you transition either into a

Scroll to top

direct federal employee job or something that I have heard before, some of the people have gone in, did the program, learned a specific skill and started their own business.

When you start your own business, as somebody who is disabled, especially for somebody if you have vision and hearing loss, that puts you into a special category and that category is called small business majority owner with disability. As long as you have 51% ownership in the company, then you can use that company to apply for federal contracts, and you kind of -- if Ability One was one of the first picks, you're second or third pick as a small business.

If you're ever trying to learn something and want to have a business opportunity to start something on your own, Ability One, Source America and those programs can kind of give you the skills and from there, you can try to start your own small business. If you go to FedBizOpps.gov, it's all on that website.

If you want to start a small business, ask me. I can point you to the right direction. Because we're focusing on federal government employment, I'm not going to do a whole other hour on small businesses because I do the contracting stuff.

So that is the end of my presentation. I hope you guys will look at trying to become a federal employee. It has been extremely beneficial for me. Like I said, we moved our whole family across the country. I'm able to support my wife and kids on just this income. And we are having a lot of fun exploring the West Coast and there are extreme opportunities for advancement. I have received three promotions since I started and I am up for another one, and they are even giving me opportunities to go overseas and work in overseas positions within my agency.

If you are ever trying to find a career field where you can have a lot of opportunities especially within the Department of Defense and want to become a leader, this is the way to go. So if there are any questions or anything like that? [APPLAUSE].

>> That was terrific, Ryan. Thank you very much. [APPLAUSE].

So unemployment is a huge issue among the community. So this is terrific stuff to know about. Federal government has a lot of employment opportunities for people with Usher syndrome. It's definitely worth looking into. We're going to take a break for about a half hour or so. We will start up precisely at a quarter to 4:00.

The next session is the most popular session here which is the USH family panel. We will see you in about a half hour or so. Thanks.

[Break]

Scroll to top

USH PANEL DISCUSSION

>> Hello, everybody. We're going to start in a minute and a half which is highly unlikely but we are going to give our best shot. We will get the panel up here and then get started.

All right. I would ask everybody to take their seats. We're going to get started in just a minute. Okay. So we are going to get started here. One of the things that I have found in this conference, it is a challenge. By the end of the day, so many people have made so many friends. Getting them back in the room is a real challenge. That's a good challenge to have. Our next session here is our USH family panel, which is usually the most popular thing that we do. And today it's going to be moderated by Monte Westerfield and Monte is a very unique person in the Usher syndrome community.

He is a researcher and a lot of the animal models that have been so critical to the research we have done, Monte has had a hand in helping to develop the models and also a father of two children with Usher syndrome which is how he got involved in this. I will let Monte take it from here.

>> Thanks very much, Mark. We have four panelists here today. Anne Schueler. Wasim Raza. I hope I pronounced that right. Okay. There we go. Anne Schueler, Wasim Raza, Sandra Scala and Jeffrey Bohrman.

And each of them I think has a very unique story to tell, so I would like them to sort of introduce themselves to you, and let's start with Jeff. So can you tell us a bit about yourself? When you were diagnosed with Usher syndrome and the type that you have, if you know where it is.

>> Oh, okay, I'm supposed to start. Hello everybody. My name is Jeffrey Bohrman and I know many people call me Dr. Bohrman. But I don't want that name. I refer to be called Jeff or young people can call me Dr. Jeff, that's fine.

My deafness was diagnosed when I was 18 months old. My mom found out that I was deaf. Before that, my mom thought I was very responsive. I would laugh and smile and make sounds. My dad was in the service.

And he would say, hi, hi. And he would say it's dada. And then I didn't respond. When I was 29, that's really when I found out that I had Usher syndrome.

I knew when I was a teenager, that I had tunnel vision. Again, when I was 29, looking back, the first thing that I can remember is riding a bicycle, and I was unable to do it because I was part of the Usher syndrome Number 1 crew, and I had a balance

Scroll to top

problem. And at that time I thought, maybe I was just a fat kid, you know, maybe that's why I couldn't ride my bike. I was a fat little boy.

Anyway, about three years ago, I had some genetic testing done. And I was in for a big surprise. I was sure that I was Usher syndrome 1F, you know, there is a high prevalence of that in Jewish people from Eastern Europe and Russia.

But I was diagnosed as 1C. And I didn't -- and that also I found out that I was French Asian and then there is a small group of people that have Usher syndrome 1C that are Jewish. So they needed more subjects or I'm sorry, there were different subtypes. And there were two groups, that and 1C.

So there is a whole long history, and I'm not sure if I have time to tell you about all that, but I'll make it short. I ended up going to the Clarke School For the Deaf and had a very strong oral program. And then I ended up going to a private school here in the Philadelphia area, Philadelphia was my old home.

That's where I grew up. I grew up in New Jersey. And I ended up going to four colleges. I have two Bachelor's Degree, one Master's Degree and one Ph.D in pharmacology.

I have been working for the federal government for ten years before they forced me to go out on disability because my eyesight was diminishing. So to make a long story short, I went to intensive rehabilitation treatment and also learned a lot more sign language, which I didn't know until I was 41 years of age.

And going back, I have to say to you, when I was 29, and I found out, my wife, we were in a new town in California where I worked to get my Ph.D and I ended up going to the eye doctor for the first time then, and he had this old report -- I'm sorry, a report from my old doctor, and then he told me that I had to go into this room as part of the research.

So I looked down at the folder, and it was opened up. And it was right in front of my face. And I was able to read it. And it said that I had Usher syndrome. And retinitis pigmentosa and I was really, really angry.

And now looking back, I really think that my parents knew since I was 10 years of age, but they never disclosed that to me, and my mom, she was gone when I was 21. And my dad was not the kind to go through that kind of thing with me.

So I don't have all the pieces of the story. My dad blamed it on my mother. And I wanted to get some DNA testing done to really find out where I was getting this 1C from.

Scroll to top

Maybe it was from my great grandmother on my mom's side of the family? And maybe it was also my great grandmother on my father's side of the family as well.

You know, maybe they were carriers and that's how I ended up getting Usher syndrome 1C so it's been quite interesting, and I changed my career, and I became program manager of statewide programming providing services for deaf-blind individuals in Ohio. And I worked there for 21 years before they ended up closing, and then I knew it was time for me to retire.

So I ended up moving with my wife to Chicago to be closer to our daughter, who also has a son. And we have a son in Colorado as well and four grandchildren.

So they are wonderful, supportive people. Oh, and also, I was the leader, you know, in local and state and also national efforts. I have been on the board of the American Association for Deaf Blind for 20 years now. And I was also on the board of the World Federation the Deaf Blind for 12 years. So I have been around the world quite a few times.

And I better stop it now. That's all. [APPLAUSE].

>> Thank you, everybody. Thank you very much. So Sandra, can you tell us a bit about yourself, how you found out that you have Usher syndrome and when that was, and a little bit about your life.

>> Hi, I'm Sandra. And I'm from Ontario, Canada. I have Usher syndrome Type 2, and I found out when I was 19 years old. I'm whatever, I'm 39 now. So 20 years later.

I basically found out through my brother. I have an older brother, 5 years older than me, and when he found out, we pretty much knew that I had to get checked out. I have been wearing hearing aids since I was 5 years old.

When I was a teenager, I noticed my vision started to get bad at night and all my friends thought it was because I had a little too much to drink. And I thought um, no. No, but in all seriousness, I still continued my life every day pretty much. But I got married, I had three girls. I have three girls. Well, now there 12, 13 and 15 years old. There was a time in my life, though, when I was working, I had avoided going to the ophthalmologist for about 10 years because I was too scared to lose my driver's license.

I knew that it was getting a little bit more risky for me to drive, and so but there was a point -- there was an incident that happened, and I just said, okay, enough is enough. It's not safe for me or my girls.

Scroll to top

By then, I had separated and was going through a divorce. So I was on my own. Anyways, I pretty much lost my license right away back in 2013/2014, just at the beginning of the year there, and I would say pretty much a nervous breakdown.

I don't know. I had gone through a lot of anxiety and a little bit of depression. And but even to this day, it's my girls that they give me hope and inspiration. And because of them, I started a blog called The Mind, Untangled and I am doing it in hopes to inspire other people who are fearful of what is to come. I always say we take one day at a time.

Yeah, so I'm remarried now. So I have three of my own. Four step children, three of which are on the spectrum and life is crazy. Life is fun. And like everybody asks me, how can you do it? Do you -- like you're superwoman. I don't look at myself as superwoman. Any of us who are going through vision loss and hearing loss, you know, it is a lot. It's not easy to come by. But I think we all learn to find the strength within us, and a within to keep moving forward so those are my reasons. They're my extended family. And even though it's not fun, it can be a couple of weeks or months, it's weird how it works, I will notice oh, the table that was right there is now all of a sudden is here beside me. That's when I notice my vision is getting worse when I'm bumping into things that are always there.

Yeah, it's scary. But I also have a K9 vision dog and she is a blessing to the family as well, so that's me. Yeah.

[APPLAUSE].

>> Thank you very much. So Wasim, can you tell us a bit about you? You're originally from Pakistan, is that right?

>> Yeah.

>> When were you diagnosed? How are you diagnosed?

>> I was diagnosed about three years ago, 2016, right here in Baltimore City in Johns Hopkins Medical. My eyes started to get worse when I was 12 years old. My deaf brother, we went to the mosque and came out of the mosque and there was a bomb blast. My brother and cousin were fine. My leg was broken. And after intensive therapy, I started to walk in after nine months, and I went to doctors to see what happened with my eyesight. It's getting worse and worse. And a different doctor, tried the glasses and after getting the glasses, I still started having a problem. I couldn't see in the dark areas in the building and definitely in the nighttime, I had problem problems. And after still going to different doctors, they're still saying the same thing. This

Scroll to top

happens because you have been in a bomb blast and there is nothing we can do about it.

So I came to the United States in 2001. And I went to another doctor. They all said the same thing. And in 2016, after 21 years, 20 years, the right doctor told me that I had Usher syndrome. And I had a genetic testing done last year and they ended up confirming Type 2. Yeah.

>> So how did you feel when you were diagnosed after all these years of --

>> I was really happy about it.

>> So it was a good thing?

>> It made me really happy. I don't know how to describe it. It was a heavy burden lifted off me that I don't have to fight a condition of looking for a cure of what happened and why it happened. But now I know why. It's Usher syndrome. And I belong to a community.

>> Thank you. [APPLAUSE].

>> So Anne, you're the mother of children with Usher syndrome so maybe if you can tell us how you discovered that and how that's affected your life.

>> Okay. My name is Anne Schueler. We are going to have our fifth kid in November, and currently two of our children 12-year-old and almost 7-year-old, both girls have Usher syndrome. We are members of the 1B club and we also have a son who is 9 and a 3-year-old daughter who are not affected.

We found out about our oldest daughter's hearing loss when she was 1 and we did not find out about Usher syndrome until our third child was born, and she also failed her hearing test and she was having a lot of the same gross motor delays that our oldest had.

And I said to the developmental pediatrician on the implant team, I said don't you think it's so weird? Their heads are floppy and doing all of the things that other babies are doing. Yeah, super weird. Our ENT surgeon said there have been a lot of advancements in genetic testing since your oldest was diagnosed. It's been five years. We should test more things. And I was like okay, I guess it's fine. And we felt really comfortable with the hearing loss.

We felt like we liked our surgeon and our kids -- we knew what school we wanted her to go to. She would go to the same school that our oldest went to. I can't say I was super interested in finding this genetic diagnosis but I kind of thought the surgeon

Scroll to top

wanted to play with his new genetic test, and I was like okay. So we did that. And I was really shocked when we got the phone call from him and he said we found the problem. Your kids, we think they have Usher syndrome. It took a little while to nail down the diagnosis because we had one known variant and then a variant that was novel. They said we don't know what this is going to do.

I did what every mom does now. And I got on Google and as soon as I read the description of kids with Usher syndrome 1B. I said oh, these are my kids. And they didn't walk until they were 20 months old. And this is what we have. So that is, let's see, Gianna was 5 and our youngest at the time, Pia was about 10 months old when we got that. So I would say that was a really hard time, especially because, you know, I think a lot of -- for a lot of us whose kids are diagnosed with hearing loss so young, you feel like you, you know, you sort all of that and you feel like okay, we have got a plan, we know what we're doing. And then somebody pulls the rug out from under you.

Now we have to learn about this other body system and how does it work, what does that mean? And what does the future hold. But I would say that, you know, at this point, living with children with this diagnosis up until now, and this is not my first time attending this sort of conference, and I don't love that they have Usher syndrome.

But I love being a part of this USH family and all of the people that we have met so. [APPLAUSE].

>> Thanks very much. We would like to open this up now to questions from the audience. We have a microphone out there. You can have mine.

>> If there are questions, just hold on a second.

>> So I have a 6-year-old with Ushers 1B and it's a big debate in our household, when do we tell her? Obviously she is aware she is deaf. We tell her she has special ears and eyes and she is already learning Braille in school. She thinks it's something fun she gets to do, gets to skip math and reading sometimes. Haha, I don't get to do that. But we haven't sat down and told her our full diagnosis and I'm wondering if any of you have advice on when to do that, or how.

>> So I would say that my advice, I mean, we're still sort of feeling it out, but that was definitely -- that's something that you know, my husband and I had been anxious over it. My husband and I, how to you talk to a 6-year-old, 10-year-old, how do you tell them?

People who found out when they were adults or older, like Jeff said he thinks his parents knew and didn't tell him ever, I don't think that's the right thing for us. But we have sort of let it evolve naturally over time. Something that has been helpful is

Scroll to top

bringing the kids to the conferences and talking to them about the reason you have your cochlear implants is because you have Usher syndrome. We're going to the Usher syndrome conference.

These are adults with Usher syndrome. You know, it's amazing how kids are really perspective but also kind of not perspective. I feel like the first few times that we came, I think the first few times we were in Boston, Gianna was 7, and I was worried she would look around and be like why do all of these people have guide dogs and these people have Usher syndrome, what does it mean for me?

She was in over her head. She thought it was so fun. And last summer when she was 11 was the first time that she kind of straight up asked me, you know, mom, am I going blind. And you know, oh, okay, here we are. She had overheard or a friend of hers, hearing kids, those darn hearing kids, had overheard me talking to her mom and then her friend, her hearing kid friend told her. I heard our moms talking and they said blind.

So we generally with our kids have tried to use really positive functional language. So we might say, you're having trouble seeing at night. We're going to make sure that you have a flashlight nearby. We're going to the zoo, it will be dark in some areas. We will be sure to have our flashlights ready. And talk about things in a functional way and the steps we're going to take to address those things.

Talking about you might have some trouble seeing to the side and up and down, we're going to work with you learning how to scan your environment so you don't trip over things as much and it has just been in the last couple of years where we started with the orientation and mobility training with the cane and even with that, sort of letting them take the lead and say, I'm making you learn these skills, they're important skills, but I'm not going to make you use your cane.

You decide. And if I notice that we are somewhere where it is a little darker or a lot of obstacles, I might say, do you want to take your cane out? And if she says, I don't need it, that's fine. But as she has gotten older, more and more she is pretty comfortable pulling it out. Sometimes I think she is just testing it out, to see what it is like.

And I also have found that I don't love to use the word blind with her because it's a very loaded word. And I think sometimes it can feel a little bit scary but also I don't feel like it tells the whole story. I told her last year, that that's not a word we usually use in our family. I want you to know the whole story. We come to these conferences because you want you to know about adults that are living full, beautiful and happy lives with your condition and I don't want you to feel limited by any of these descriptions.

Scroll to top

And I just think it's important to keep an open mind with communication with our kids. Say, I don't want you to worry about this. You might worry about it. But I want you to be able to talk to me and you know, if you have a problem at school or a problem at home, like we can figure that out. There are so many technologies that we have available, and all of the doctors that are working on so much research. There are so many things to be hopeful about.

So I think that talking about all those things with your kids in an age-appropriate way, it will be a continuing conversation. I don't know that you have to necessarily plan this one sit down moment and be like okay, we're dropping the boom. I think it can kind of, because it's a progressive disease, their information and their understanding can progress along with that.

You know, I know it's hard to watch our kids suffer and I think it's okay for us to have days that we feel sad and we grieve and our grief can come and go. Things change. You get used to the way things are. It changes again and you have readjust and I think letting yourself do that is really important so you can be supportive for them and, you know, not make it feel like it's this secret thing.

I feel like you know, when she was first diagnosed, a lot of other people here would say, you know, they're still the same kid. They're still the same kid as they were before you knew they had Usher syndrome. Anybody have anything else to add?

>> Yeah I think Jeff had something to say.

>> So one thing I would like to talk about is the use of a cane. When I was around 40, I started to use a cane. I didn't use it that much, but now I do. It's a supportive cane. Because I have balance problems. That aside, I am very thankful to my mother who was very strong and independent herself.

She would drive me place-to-place. I never learned to drive. One thing looking back, that really scares me now, is that my mom would encourage me to go to New York, and that started when I was in high school. There was a club every Friday night where we would have social time, and I would go to the train station after school and take the train to New York, and then I would get some dinner, and I would take the subway over to the club.

Around midnight, I would take the subway back and if I knew about having Usher syndrome at that time, maybe I would have been more fearful to go to New York. I can't believe that now. I was very, very independent even at night. So maybe it was the right time and maybe it was good that my mom didn't tell me. I don't know.

>> More questions?

Scroll to top

>> Well I think he kind of answered the question. I was going to ask you, Jeff, about how you felt about your parents when you found out that they had known and perhaps not told you.

>> Well, I really do wish my parents had told me before I went to college because I picked the wrong career. And my dad would say, okay. Then what did you want to do? I said, well, I plan to become a teacher. I was going to go to Columbia University in New York to get my Master's Degree and go get my doctorate in educational administration so I can run a school system.

Or become, you know, a computer expert. But my dad said that he wouldn't support me in going for that education to get my Master's. And I love medicine and I wanted to become a doctor.

So I decided to go to school of pharmacy. But that was also a "mistake" you know, I really wish I knew this. It is very important that you have a more informed career plan.

>> Before I take the next question, I wanted to add my own two cents as a father. Try to talk to your children as young as they can about the fact that they have this disease. As Anne said, they don't get it. It just goes over their head but it just starts to become who they are. And if you talk to them a little bit about it, you better make sure to bring a flashlight with you, it's going to be dark and you have Usher syndrome, and that's who they are.

It doesn't make it as big of a deal and they don't sort of have that crushing moment when they start to understand what is going on and it just becomes kind of who they are for as long as they are. So that's my advice.

[APPLAUSE].

Next question.

>> Hi, my name is Dianne and my twin boys were just diagnosed at Christmas. They're 21 and have one more year at college so I kind of have a couple of questions for you. One is, when should they -- they will be job hunting in the next spring. When should they be exposing this to their employers? Right now they have very functional vision but we know it's going to progress. And the other part, how do you know when you're done driving without crashing? And then I'm kind of looking for some guidance on our responsibility with sharing with family.

Like my immediate family we know, but what about cousins and things like that. So those are my three questions.

Scroll to top

>> Sandra, do you want to grab this one?

>> Can you start from the first question again?

>> So when would you suggest to tell employers about this, that you have Usher syndrome?

>> When did I find out?

>> When would you tell an employer that you have Usher syndrome?

>> Oh, maybe I'm the wrong person to ask.

>> And then the other question which you are maybe more comfortable answering. When do you suggest you stop driving beyond a crash, and learning that Usher syndrome was the cause of the crashing.

>> Yeah, I know, right. Those are good questions. They are.

So really, honestly, I didn't know so I didn't know I had Usher syndrome until I was 19. I honestly -- so I don't know. The laws are very different between Canada and the United States. Obviously every country is different. But I didn't tell my company right away because I was scared. I really was. I was really scared.

But I was with that company for 15 years. Eventually when I got the final diagnosis, the official diagnosis, I did tell them. And they kept me on, and they treated me equally. I have no grudges against them because I felt better telling them. My vision started getting worse and people knew if I needed something. They would put things out of my way so I didn't bump into them. And that being said, too, there is one day there that I bumped my eye into the printer tray, that was sticking out and I ended up bumping my eye and I ended up having to go downtown to get stitches. I was Downtown Toronto.

And another time I got hit by a car. And all these things, that's what added up to me not being able to work anymore and my anxiety took over but if I had the chance and more accessibility options like if I had more knowledge at the time, I would have said to somebody from work, hey, can you walk me down to the station so I can get to the train so feel comfortable working in a very busy area.

As far as the license, like I said, I avoided it as far as I could but my whole thing was I didn't want to put my kids in danger. And then my brother continued to drive even after I lost my license, and I felt uncomfortable driving with him. It got to that point. You get a feeling what you don't want to put others in danger and you don't want to put yourself in danger so what is the right thing to do? Well think of all of the options and then if you're in a suburban area and have no means of getting around, you have to look for

Scroll to top

support to help get you around. At the end of the day, it's to help keep you safe and others safe.

And I think a lot of it was, I was, I was embarrassed at the time. Embarrassed to admit to people, yeah, I'm losing my vision. I lost my hearing. It's what makes you different. You are who you are. Everybody has their own issues and you have to figure out how to make it work for you, and your family and the people you love. Yeah. Is that okay?

[APPLAUSE].

>> Question down here.

>> Hi, my name is Fran and I graduated from Teacher's College, got a Master's and a doctorate. Great experience. If you had not gotten those experiences, you may have been another way, but you got a lot of great experiences. Coming backwards from the educational point of view, and I had an opportunity this afternoon, the very first baby in my infant program was a Type 1 Usher syndrome and I think the thing that made him the most incredibly intelligent and brightest kids -- young men that I had ever worked with, was the fact that he signed. Got a cochlear later on. It just wasn't going to work.

His mother learned along with him. She never gave up and whatever he was doing, she kept on doing. For those who use sign language, not the worst thing in the world. For those that do sign or in the auditory world, you have got to keep up with your child and I found this young man was successful to the point where he went to Gallaudet for undergraduate and took all his courses at Georgetown with interpreters and ended up going to Stamford University and passing the bar in New York and California.

I truly believe it was because his mom learned sign language. Not the first 50 words or the little book. What I see here are an amazing amount of parents, moms and dad, who are so supportive. It's not just now when your children are nice and little and cute, but forever. I tell everyone, you parents are the children's advocates forever. Your sons just got it at 21, but you're always there. Even up in heaven, I still hear her voice. You guys are there for the children. I know I'm talking too much. You guys are inspiring on the stage and every parent here be there for your child, whenever you want to tell your child, whatever level. Be able to communicate. Give them access to communication from you, whatever school they're going to go to and wherever they're at, be sure that they understand people communicating.

[APPLAUSE].

>> Thank you. Are there other questions around here? A question in the back here.

>> Hi. My name is Aaron. First I just want to say, everyone who is sitting up on the panel, you guys are incredibly inspiring. My eyes and ears were glued to the stories

Scroll to top

that you were telling. It was super fascinating and it really seems that you all live full and extensive lives, even with the disabilities that you have.

And my question is with the full and extensive lives that you have, transportation and mobilities is one of the biggest challenges that we face with Usher syndrome. Can you focus a little on how you do what you do every day and make such great impacts, get around and all that stuff and deal with transportation and mobility?

>> So the question was how do we deal with transportation and mobility as Usher people?

>> In the daytime, I take the local bus by myself, and I also have the disability service if I want to go further somewhere, I can order that, or take a look at taxi for the lower fare but in the evening time, I have more difficulty. I use Lyft sometimes and of course my wife most of the time take me wherever I want to go.

>> Okay, so I will just touch on the time when I first lost my license and so where I was working, there is offices both -- I don't know if you're familiar with Ontario area, there is Mississauga and Ontario. I had to get transferred to the office. It was going to get too expensive. And I ended up getting transferred to Toronto. When talking about transportation, you're going to do what you got to do to survive kind of thing, right? You somehow find a way. For me I would walk every day. I would walk 45 minutes to the station, grab the train, walk another 10 minutes at night. And then pick up the girls and carry on. Was it easy? No. You get yourself into a routine and find a way to do it and there is like I know, even in the small town we have now, there is mobility things. So for people with disabilities, they don't drive and whatnot, you know, they provide transportation for you where you have to go, go get the groceries and whatever you may need. Right. It's just a matter of looking into your resources to find out what the best mode of transportation would be for you. Yeah.

>> Jeff, do you want to answer that question on transportation mobility?

>> Okay. My wife and I, we got engaged. That was 49 years ago. My dad asked me, if I could explain to my wife about my eyes. And I said yes, I had tunnel vision and I'm unable to drive. But what my father really meant is that I should tell my wife that my vision was going to get worse and worse every time.

And I asked my wife if she knew, you know, if she wanted to continue to marry me. And it was quite awkward, but we had a wonderful marriage and she was a really good support to me. And also in the '70s, my parents told me that I couldn't drive.

Scroll to top

They said, they thought I would get married and didn't think I would have children. They told me not to have children. My wife got pregnant with our first child and my dad was jumping up and down, so there you go. [APPLAUSE].

And I think my parents were fearful that I would pash my Usher syndrome onto my children, and my kids know that they're carriers. And my grandchildren are carriers as well. I didn't really tell my grandchildren yet. I just said to my children, like when they were 12 or 13, my daughter asked, you know -- she started to cry because she thought she would become blind herself. And I thought no, no, no, that's not the case. I tried to explain to her that there are people like me that are deaf and can become blind. But that's not her.

And I had a son, who is hard of hearing just like my wife, and the eye doctor that my family goes to was really ignorant. When he saw my son with a hearing aid, he said oh, well he has to have an ERG test done. Oh, my.

So there are a lot of professionals that need more education. Now they are more educated and there are still some of them out there that really don't fully understand, so we need to teach them. So are there other questions? There is one over here.

>> Hello. I wanted to ask about diet. Sometimes I find if my son has too much sugar, his balance will be affected more or doesn't get enough sleep. I'm wondering if anybody eats junk food or stays up too late, does that affect your balance? Or worse days. Not feeling so good today and your eyes are worse some days than others or is it pretty consistent.

>> So the question was about diet and sleep.

>> Well I think you know there is generally no question, right, that we all function with the correct amount of sleep and eating healthy things. I know that I'm going to make every mom here feel really adequate. Sometimes my kids eat really well. Sometimes they don't. Sometimes we get them to bed at a good time and sometimes we don't. If I am feeling really hard core, might research good DHA supplements and push them on it, and then I realize it has been two months and no one has taken a vitamin of any kind, and I think that, you know, it's hard for me to sort out I guess when I look at my kids.

Are you acting like a lunatic because you are overtired or just kind of a lunatic sometimes?

And I think that diet and making sure that they are sleeping well, I think these are good habits for them to have throughout their life and it certainly doesn't help and I also think

Scroll to top

that, you know, there isn't a whole lot that we can do as parents and there are times that I might say, you have to wear your sunglasses if you go outside. You have to.

I cannot -- you know, I'm not going to be studying oligonucleotides, but I can make you wear your sunglasses. You are definitely going to do that. As a parent, I think my kids function better if they get X amount of sleep or eat this thing or don't eat that thing, I think that can alleviate a lot of our own anxiety to feel like I am doing something positive when there is really not a whole lot in actuality that we can control. But I also just want to, I don't know, I don't want anyone to beat themselves up or feel guilty about I should be doing this, should be doing more or you know, nobody should have to live like that. We have enough guilt as parents. Be gentle with yourselves, and if you're up for like serving them liver or whatever and want to fight that battle, you should totally do it.

If you feel like we're just going to have some mac and cheese tonight, give yourself permission to do that, too. But yeah, I don't know. The other panelists have noticed personally as adults, being able to draw the distinction, do you feel better eating certain things or getting sleep? But yeah, that has just been our experience.

>> I think we have Wasim's office.

>> It can make me feel more tired. And take it out of me every day. Constantly scanning. Right now I have difficulty with echo sounds in the background. I am glad to have that offered but it takes energy out of me. More veggie, more fruits. But I know my wife tries to get me to eat that most times.

>> Okay I have time for one more question I think. And I'm going to have to skip you guys because there was a question back here.

>> All right, so I have Ushers Type 2 and the biggest thing is trying to figure out the coping of the progressive, how to deal with the change -- how you guys handle the coping cycles we go through and the anxieties we get. That's the battle that I am dealing with right now and I was wondering if you have any ideas, advice or tips or whatever.

That is a great question. So if you didn't hear the question, do you want to repeat it?

>> So the question was how do you deal with the continuing loss of vision? So you learn that you are losing your vision, deal with that, everything is okay and then it gets worse and that's a continual cycle of coping and she is asking for advice.

How do you -- how can you manage that better?

Scroll to top

>> Just don't lock yourself up in the bedroom. It's not a good thing. Its taken -- it is. It's hard. Right. It's hard because you feel like you're losing your independence. You feel like there is just a part of you, like you just feel lost. And you want to talk to people about it but then you don't want to be a burden on those other people and you try to deal with it yourself in your head and there are a lot of people that do meditation and you look to a higher source or whatever, right.

Everybody has different beliefs. And I think the main thing is, is that in our heads we already know that we're going to be losing our vision, and it may get worse. We don't know how fast it's going to progress or how slow and we know that we have people like Usher Syndrome Coalition and different organizations trying to make a go of it.

You know, I think it's just a matter of just hanging on. You just got to find something that makes you happy. And know in your head that there are people who love you, they support you and you're not alone. There is a whole room of people here that holy moly, I had no idea that there was a room that existed of all of these people coming together like this. It's really amazing.

I called my husband up. Holy smokes. There are a lot of people here and I want to get to know everybody to see if they feel the same way I do. It is an ongoing challenge and you know, life is too short. Life is too short. Easier for me to say. Easier said than done. We will all find a way to help us keep moving forward. Deep breaths. It goes a long way. It helps. It does. That's what I find. Yeah, and trust me, I have my moments where I cry and ask myself why, and you know, my parents have felt guilty and whatnot. They think it's their fault. It is nobody's fault. That's what we have to realize. As much as we have our issues, there are people who have their issues as well and everybody finds a way to hope in their own way.

You got it. Don't worry. We all have the strength to do it I think. We do.

>> All right. Going through my vision loss has been very, very difficult and very painful and what has really helped me is that my father taught me that there is always solutions to every problem that I would encounter. In life. And that really helped me to keep going and to tell you the truth, I'm very, very happy to be at the end of a very long road.

And then because I just finally became fully blind. It was very hard going through different stages, but I'm very impressed with all of these young people here.

And I know that my parents, they knew back in the 1940s when they got me, and they were very, very thankful. And I was thankful to them.

Scroll to top

But I couldn't understand speech. And that's one of the things that I think that the cochlear implant is important for some children that are deaf. And that's my own opinion. Because some of you might think it's best to be a part of deaf culture on the other hand and I hang out with deaf people, and we use ASL. I learned it late in life. But the general point, going through the different stages of vision loss, is very hard. Thank you.

[APPLAUSE].

Wasim or Anne, do you guys want to answer this? How do you deal with the changes, Anne with your kids and Wasim, with the changes you're dealing with personally.

>> I think I guess I -- my perspective, you know, I think for me, just part of reminding myself that because things change, it's okay for my feelings about to change. It's okay. And to -- if you have a support system around you of people you're either really close to and may not be experiencing the same things and you feel comfortable being brutally honest, man, I am having a tough day today for whatever reason or meeting people here who really know what it's like to have and watch your kids go through something like this, I find it really liberating just to say, this is an ongoing cycle of grief and finding healthy ways to live with it, live alongside it.

>> I had no support in those days. I was the only one. There was no group. Remember, I'm a little bit older. But it is very important to have a support group.

>> Yeah, I can't imagine, you know, doing this alone for sure. And I think that for our kids, teaching them the skills of how to cope and like what Jeff said, every problem has a solution. Telling our children, when we come up against a problem or if you come home and say I had a really hard time with this thing at school or I am feeling sad about this, okay, let's talk about it. What can we do. And you start running through what if this, what if that questions.

What if? What if my kid never drives? Well, I don't know. I hear a lot about these self-driving cars so that might be a thing.

I think that there is a lot to be positive about. And it can help to channel that fear into saying okay, I'm feeling sad and afraid. But what am I really going to do about it instead of letting it paralyzing you. Or finding healthy ways, going for a run, swimming, I like to knit. Finding those things that bring you joy can be helpful in alleviating some of the really acute moments of anxiety.

>> I know it's 2019 and everybody wants everything fast, fast. You can take your time but it doesn't mean you can't be slow. To get on time, you just get up earlier. You can still do everything, just take your time. It's possible. That's all. [APPLAUSE].

Scroll to top

>> Excellent. Excellent. Thank you, guys. It was fantastic. We appreciate you sharing all of that stuff. One more round of applause for you guys. Thank you very much. [APPLAUSE].

Scroll to top

CLOSING

>> Okay, if you look to your right, you will see people that came in. Are Gary and Ethan in there somewhere? Are these Gavin and Ethan. They have spent their day here in Philadelphia. And they're going to come here and tell you about it.

>> Hi, everyone, my name is Ethan. Today, me and Gavin and Ava spent the day with children with Usher syndrome, a child who has Usher syndrome and all day we had fun making sure we got to know these kids and understood they were in an environment where they were understood. I have Usher syndrome, and I have Usher syndrome, too. I will let Ava speak.

>> Thank you. It was really amazing to see how we all connected no matter where we were from or what we like to do, we were all connected. It was just so amazing to see the smiles on everyone's faces between everyone. An amazing feeling. You can just feel the energy in the room.

>> And another reason we do this camp is so that we can all bond together over this and we can all feel comfortable with each other, as well as being connected through different activities. We did several different activities where we expressed something about ourselves and showed how other people are connected because they like the same things and also showed how we can raise awareness as people with Usher syndrome by telling people what it is and how they can help people, if they know someone who maybe has Usher syndrome but not even if they have Usher syndrome. They can be deaf or have some visual impairment but it's just good to get it out there so they can help if they're in that situation where they need that.

[APPLAUSE].

>> So we wanted to just share today. This is kind of a little example of some of the opportunities that we like to share as an organization at Ava's Voice. Opportunities for families in May of 2020 for families to get together, engage in resources and our big goal is to share about the USH this summer youth sleep away camp. And it's a youth camp. And today we have gotten feedback from the kids on what they like to do and hopefully we will get better and better as we evolve and grow and we would love your feedback and for you to be engaged with the organization.

I'm going to have my youngest daughter Miley here who does not have Usher syndrome, but she is the best little sister ever and she is going to pick our winner.

Some of you shared contact information at our table. So Hunter is the winner of our Echo Dot today. [APPLAUSE].

Scroll to top

Hunter, can you can over? Do you just want to share with everybody, did you like camp today?

>> It was perfect.

>> And would you like to come to our summer youth camp do you think in the future?

>> Well it's in New York, yeah, I would love to come.

>> All right. We're going to put you on the VIP list. Thank you, everyone. We appreciate it. [APPLAUSE].

>> Mark Dunning: Okay. Thanks, guys. I hate following people like that. So this is it. Thank you for coming today. Oh, the pen that we have been passing back and forth all day long. Thank you for the pen, whoever gave us the pen. Thank you.

So thank you all for coming. This was a fantastic event. You know, I'm not saying that because I want to take credit for anything. Having you guys in my life, makes my life very worthwhile. And you know, there was talk about coping earlier. This is how I cope, is with you guys. And I hope you find it to be the same and I hope that these kids are able to experience the same thing. So this is it for this year.

Next year we have been batting around ideas. Should I tell people where we are thinking of going? Okay. She is nodding. We are thinking of going to Austin, Texas next year.

[APPLAUSE].

So there will be more information to come about that and where it is and how we will organize it. The good news is that we're growing, as you can see by the size of the room. The bad news is that is starting to limit our options on where we can go because there are so many of us. It's a good problem to have. There will be more information to come about that and for those of you who have got tickets to the social, that is at 6:00, so you get an hour break here. And it's in the room where we had our lunch.

If you did not get tickets to the social, Krista, you can get tickets at the door if you did not get tickets to the social. She was trying to avoid that, but tough. If you do have tickets, it's next door. If you want to come and you didn't get tickets, it's next door and we will figure out how to get you in. So thanks, everybody, and we will see you next year in Austin. [Applause]

Scroll to top

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Scroll to top