Transcripts of USH Connect 2024

Morning Presentations: 9:30am-12:30pm

Carol Brill, Welcome Speech
Jacqueline Turner Presentation
Dr Conan Donnelly Presentation
Dr Solomon Kamal-Uddin Presentation
Kidist Puig-Lynch
'Are you Usher Aware?' Launch

CAROL BRILL, Welcome Speech:

Hi everybody. You're all very, very welcome. I just want to say thank you for all being here. This is a very special day. It is international Usher Syndrome Awareness Day. There are people all over the world celebrating and let's hope we feel their vibes around the world. I'd just like to say thank you to all the volunteers for just helping to make this event happen. In the run-up, a lot of work and preparation went in to organising this day. I'd like to say thank you to all our sponsors and thanks in advance to our speakers. You're very welcome. We're looking forward to hearing what you have to say. We want to thank our accessibility support, which is Michael, Marea, and also Claire of the National Museum of Ireland and to the Irish United Nations Veterans Association. I want to say thank you to the board of directors, Deborah, Anne, Trudy, Deirdre, David and Eugene. What we have achieved so much to date as a charity in just under three years. It is quite a lot, but yet we have more goals to focus on and for that, we need help. We need help from you or if you know somebody that can help us that would share their skills with us to help achieve the goals on your behalf. This is your charity. We would also welcome fund-raising ideas or indeed, if you know of anyone thinking of nominating a charity, please tell them about Usher Syndrome Ireland. Currently we have two support groups. One for parents. This is a compliment to the great work of Usher Kids UK. The purpose of the parent group is to focus on Irish issues. We also have a peer support group for cochlear implant users. As I said, this is your charity.

Today our event is called USH Connect and the word, "Connect" is very, very powerful. Just ten years ago I met Deborah at a conference for the first time. We had connected through Twitter and little did I know that Deborah and I would be working together ten years later and fulfilling a dream that is Usher Syndrome Ireland. So thank you Deborah for treating me and I won't say anymore! Deborah introduced me to our fellow director Anne Shields and we had an amazing evening in Derry ten years ago. What we have gained together will go from strength to strength because the connections that you have made already or the connections you have yet to make today is something that will grow into something beautiful. The importance of connecting with someone who shares the same Usher journey is a very important part of our wellbeing. I can speak personally from the bottom of my heart that I have felt so lucky to have the friendship and support of the Usher community this year especially. Some of you may remember that last year at our Usher event I was not in great shape because my hearing was deteriorating quite rapidly. So last April I got a cochlear implant and while it has been a bit tough, it was good to be able to lean on my Usher friends for support for answering my silly questions and for cheering me on when times are challenging. So that's at the heart of Usher Syndrome Ireland. To support each other and I have to say my heartfelt thanks to Deborah for connecting me with so many people over the last ten years. She has been a driving force and you can all see the hard work that she has done for this charity for you. Thank you, Deborah. (APPLAUSE) So now we have a great day planned ahead and I hope you will all enjoy it. I'm going to pass back to Katie to introduce our next speaker. So, have a wonderful day everybody. Thank you. (APPLAUSE)

JACQUELINE TURNER PRESENTATION:

I'd like to say it is really wonderful to be here today. I met Carol sometime ago now, but I think Usher Syndrome Ireland is a wonderful organisation and it is a real pleasure to have been invited to speak to you today. Charles Harod discovered or Delineated Usher Syndrome nearly 100 years ago. I'm going to give you an update on the clinical trials, but I want to discuss the progress that's been happening on rare disease initiatives in Ireland because I think that's very important for all rare diseases and Usher is a rare disease. There are over 6,000 rare diseases and Usher Syndrome is one of those. So, I work in the Mater Hospital and I'm a Genetic Counsellor. For the last couple of years I've been working oven the Target 5,000 study.

It was a study run by Fighting Blindness, they realised it was very important to have a genetic diagnosis. So Usher Syndrome is cause by over 12 different genes. It is important to have that diagnosis. First of all to confirm that it is Usher Syndrome that you have. You don't have any other form of deafblindness which is possible and secondly you know what the risks are for having a child with Usher Syndrome. So because it is a recessive condition, that would be very unusual. So you wouldn't expect to have a child with Usher Syndrome if you have Usher Syndrome yourself. So, the Target 5,000 study is a three-pronged study. We do genetic analysis first of all, we want to manage patients. So we want to see you every year or two years. Usually every two years. We want to make sure that you have your best possible vision and that's very important to us. Then the third aspect of the study is that we put your coded details on to our register. So on to a register. The Target 5,000 register, but we're actually going to change it to a different register this year which will be incorporated into the European register. So we have access to European data and they'll have access to our data. So, this slide is really about, you know, Usher Syndrome and the effects that Usher Syndrome has and you're all very familiar with that, but we can still classify Usher Syndrome into four different types. Basically, you can say that Usher Syndrome Type 1 is the most severe. Children with Usher Syndrome Type 1, they are born profoundly or severely deaf. Now, they don't know they have Usher Syndrome at that same because at that stage they think they've got deafness, but they don't realise in time they're going to start having problems with their vision. So Usher Syndrome Type 1 is the most severe. Usher Syndrome Type 2 is the most common. For children with Usher Syndrome Type 2, they have mild to moderate hearing loss at birth and they will develop retinitis later on. Usher Syndrome Type 3 would be quite rare. We only have a few families that we're aware of that have Usher Syndrome Type 3, but it is a very common condition in Finland. So, in Finland up to 40% of their Usher patients are Usher Syndrome type 3, it is a studied type even though it is quite rare. Then you have the Usher Syndrome Type 4. They're very rare and it causes late on-set hearing loss. Now, balance is an issue for patients or for people who have Usher Syndrome Type 1. Usually they don't realise because at this time they think, "I've got hearing loss." They think the hearing loss is causing problems with the balance. So they can have delay in their walking and they can be described as quite clumsy in walking and there are certain

activities that they would find a little bit more difficult than other people because of these balance issues. Some of the research says that people with Usher Syndrome Type 2 sometimes suffer with balance issues and certainly some people with Usher Syndrome Type 2 have told me that, that they've had issues with their balance and there is a study that says that people with Usher Syndrome Type 2 can have asymptomatic loss. They can have issues with their balance, but it is not really obvious. When I looked at the studies, it said 40% to 80%, but when I looked at the study, the 80% was four out of five patients. It was a tiny study, but you have to be careful when you're reading the literature because the percentages always mask the numbers. So you need to go to the study and see how many numbers did that mean? It is important to look at the studies and see how many patients or how many people were on that study. Type 3, you can have variable balance issues. The visual loss is what we deal with in the Mater Hospital and the visual loss for Usher Syndrome as all know is retinitis pigmentosa. Type 2 and Type 3, it occurs in the teens or maybe early 20s and Type 4 later on. It is progress retinitis pigmentosa. From one family to another you can have variations. These are just ballpark times, but from one family to another you can have variable changes in how they present and how severe the condition is. So how many people in Ireland have Usher Syndrome? We don't know. So, there is no study at the moment that tells us how many people have Usher Syndrome in Ireland. So like all rare diseases we don't have a rare disease register. A couple of years ago I started working with the National Rare Disease Office. I thought I'm going to start a rare disease register. It is much more complicated than you think. But estimations worldwide is there are between four and 17 people per 100,000 people with Usher Syndrome. If you look at Ireland, there is a population of 5.1 million in Ireland and that will mean 204 people in Ireland with Usher Syndrome. Now we suspect that it is much more than that. So, it is not the four per 100,000. It might be in the middle to the 17 per 100,000. If you look at non-syndromic hearing loss. The number of people with that is one in 1,000 people and that's caused by over 100 genes. Retinitis pigmentosa is caused by 80 genes. Usher Syndrome is caused by 12 genes. So, Usher Syndrome accounts for 18% of all retinitis pigmentosa and 3% to 13% of people are congenitally deaf. It is so important that we actually clarify the genes that are involved and we check the diagnosis. That's the whole idea of Target 5,000 and for conditions like this that we're

clarifying exactly the diagnosis. If there is no rare disease registry, you cannot decide how many people are in the country with the condition. You can't provide for it. You can't lobby the Government and say, "There is all these people with Usher" because you don't know the numbers. You don't know where those people are based. One of the reasons for the Target 5,000, we have a register. So we have a register so we can say to companies we have this number of patients or people with this condition. So, I'm not going to go into this. This is a busy slide, but it is saying that there are six genes that we know that cause Usher Type 1. Three that cause Usher Type 2. For 10% to 15% of people with Usher Type 1, we can't find the gene. The test that we do at the moment to look for the gene. We look at when a gene is being coded - there are coded regions which are made into protein. So a gene makes a protein. It is a recipe that makes a protein. There is a code that turns into the protein from the gene and there is also code within the gene that doesn't get turned into protein. So, sometimes people have mistakes in their areas that don't get turned into protein, but it affects how the gene is being read. So, you need a better test to test those people. So the ten to fifteen percent, we need a better test to find the genes that are involved. Sometimes we're pretty sure they have Usher Syndrome. They have signs of Usher Syndrome. We might find one gene change, but have not found the match. We all have two of every gene and therefore, two Usher Syndrome genes. Sometimes you get a missense variant. A missense variant means that the genes that cause Usher Syndrome make proteins. Proteins are made from amino acids. The wrong amino acid has been put in at some point. Usually they cause a milder condition. People with Type 1, they might have a sense variant and they might have some residual protein. We know there are some Usher Type 1 genes that only cause hearing loss and never turn into Usher Syndrome and the same genes, but different mistakes on them can cause Usher Syndrome because they affect both your eyes and your ears. The same is for Usher Type 2. We have a number of people with Usher Type 2 that have RP only. They don't have any problems with their hearing. They only have problems with their RP. They have missense variants often. If we look - I don't know if you are aware, but there was a study from Target 5,000 on Irish patients with Usher Syndrome. This is the sort of things you can do when you have a register. That's why registers are very important. You can actually work out where are your patients. Where are they coming from? So, if you look here at the

first graph, about a quarter of people in Ireland have Usher Syndrome Type 1. Okay. So we have severe Usher Syndrome. About three-quarters of patients in Ireland have Usher Syndrome Type 2. Then if you look at the other graphs, you can see that the vast majority, so this one here, let's see if I can work this pointer. The middle one. The vast majority of people with Usher Syndrome Type 1 have the gene that is involved in Usher Syndrome Type 1. The vast majority. For Usher Syndrome Type 2, the vast majority of people have Usher Syndrome Usher 2A and the next biggest one is ADGRB1. If you look at those two genes, they represent 80% people of in Ireland that we are aware. There are 145 people on this study and if you take that, the cost of illness study which estimated the number of people in Ireland with Usher Syndrome and that's where we got the 76% from. But we're missing people. We're missing people with Usher. So we want the full picture. It is very important that we get the full picture. What do they do? What do the genes do? So, all of these genes make proteins. These proteins interact with one another and. So they form a complex and if they're missing, you will run into problems with your hearing and photoreceptors and it affects the cochlear. Sometimes Usher Syndrome is called sillaophy. They can be divided into a mobile hair-like projections which move and non-mobile. The photoreceptors are examples of non-mobile. This one here is a picture of the cochlear, the blue and the red picture. This is the trachea. We have these little finger-like things that move things around. We have them in our trachea and our digestive tract and our fallopian tubes to bring the eggs to the womb. The tails are sillia. This one at the end is a picture of the photoreceptor. I'm going to talk about that in a minute. Where are the genes located? This is a photoreceptor here. They're badly ringed by myself! These are all working within the photoreceptor. So for Usher Syndrome, the genes that make the proteins, they work inside the photoreceptor. They worked in this area here. Hold on. I'll go back. They work in this area here. What do they do? This area here is called the outer segment. All these little things here are your disks. They're what we use to absorb light. That's what the photoreceptor uses to absorb light. So the complex, the proteins that Usher Syndrome all collect in the area where proteins, billions of proteins are going in and out of that outer segment all the time. Although we're born with a certain number of photoreceptors and we don't make photoreceptors as we go through life. But what happens is, the very tip of that outer segment gets worn away and every day we replace

it and the full outer segment disks get replaced every 10 to 12 days. If you've got a problem with proteins going in and out, you're going to upset the balance and that's going to upset that outer segment and without the outer segment, you won't be able to see. How do we manage Usher Syndrome? You're all very familiar with how we manage the hearing side of things. We try and ensure that people, if they have any hearing, that they get a hearing aid as soon as possible. Then you have the cochlear implants. Speech therapy is very important too and sign language. What we would like is for people to come for annual auditory examinations. They can check if you're completely deaf. They can check your ear drum to ensure it is working properly so there is no infection. There is no infection behind the ear. So that's important. And then the next bit is what happens in the eyes. You're probably all familiar with these pictures. I'm just going to go through what they are. The first is picture which is looking at the back of the eye. That's the image of the back of your eye. That's the first picture. We'll take a photo of the back of your eye just to see that everything looks like it is working normally. For this, you expect a lot of pink. You see that area where it is dark in the middle, that's the central vision. The white air is the optic nerve. That's a normal picture. The next one beside it is somebody with retinitis pigmentosa. You use a dye. You take a picture of the retina when it is being dyed and you expect a light grey. If you see dark areas, the dark areas usually indicate that the cells have died. So dark isn't good. If you see bright areas, that means the cells the struggling. This picture here shows you the very dark dot is your optic nerve, but around it, you can see the dark areas where you have cell death. The next picture, you can't really see it, but it is a picture of the visual field. I'll just show you. Here. I'm sure you are all familiar with that, but that's the visual field is where you check to see what your side vision is like. You always expect a black dot in that because that's your optic nerve. If you have retinitis pigmentosa, there is a dark area. If you look at your own image when you're next in the hospital, they'll show it to you and you can see the areas where you're having problems with your vision. The next is reading the letters on the chart. For normal vision, you're suspected to read the second last line. The next picture is OTC, Optical Coherent Tomography. If you have problems with your eye, you might have a little cyst and you will see that or your retina may be coming away and you can see that on that imaging. When you have problems with your retina, the layers get thinner. So you can also ask can

you see your pictures. The layers get thinner. Sometimes there is area which is where our photoreceptors are and that can get smaller when your photoreceptors are in trouble. The next picture is just showing what a normal eye's response would be to a light stimulus. You might have heard about a negative. That's when your photoreceptors don't respond. You can have problems with your balance. Physios can help with desensitisation. There is a type of therapy which desensitise you. We looked at the cochlear implant. There was a review of 33 studies done for cochlear implants. Again, you're looking, there is bias here, okay. Even if this review with all of the papers that they saw, 56, most of the people in those papers have Usher Syndrome Type 1. Only nine patients with Usher Syndrome Type 2. Generally cochlear is a good thing. It seems to be beneficial. It improves speech. It improves quality of life. It improves independence. So, most people generally feel that cochlear was a good thing when they had it done. Now, they didn't all use the same measure. So that's another issue, but there is hope that in the future, they will use the same measures of quality of life etcetera so that we all are doing the same thing. Basically, most people find it a good thing. There were some reports of a negative effect and some people felt there was no improvement, but generally it was a good thing. I think this is where it is important to screen children who are congenitally deaf. If they're going to run into problems with their vision in the future, maybe the cochlear implant should be something that we should have rather than using hearing aids. So that's another thing to think about. What are the treatable conditions? I said we like to see you every year or two years. We say two years, but usually yearly for conditions where your retina might come away. That's not Usher. But every two years we want to see you. We want to see you because if you've got Usher Syndrome, you've got a higher incidence of cataract. There is a higher chance that you develop cataract. You've also got a higher chance of developing like a fluid within the layers of your retina. Usually in that area that you need for clear vision. So quite suddenly, you realise you can't see - you're seeing through a fog. It happens quickly. So when we looked at the study last year. They found that in Irish patients, 77% of patients with Usher Syndrome had cataract. In most of them, they were by lateral cataracts and a quart of our patients had this oedema. If you have a cataract surgery, you might be at increased incidence. Kirk didn't find there was an association. I'm just going to talk briefly about clinical trials. There are so

many clinical trials. Generally if you look, so there are two websites there. One of them is called clinicaltrials.gov

You can put in Usher Syndrome on the Clinical Trials website and they'll show you all the studies that have been done on Usher Syndrome. Natural History Studies and sometimes population studies. They're looking to see what is the genetic makeup of the population. So something like the Target 5,000 like we've done. Then the gene therapies are all in there. Then there are gene independent therapies. Therapies applicable to all people with retinitis pigmentosa so it doesn't matter where your gene is and over the counter treatment I'll quickly mention. The key factors for hindering progress with Usher Syndrome gene therapy. The main one is they actually don't know what these genes do. So, there is a problem with basic science. They know where they are. They know where they're located. They know they interact with one another, but they don't really know what they do. You don't need a lot of these proteins in your photoreceptors. That's a good thing, but how do you get this protein into your photoreceptors? A lot of the retinitis pigmentosa, the genes work with the retinal pigment. The layer under the photoreceptors. That applies to more than one photoreceptor. If you can get something in, it will supply more than one protein receptor, but for Usher Syndrome, the genes are working within the photoreceptor. So we need to get the genes, a working copy of the gene into every photoreceptor. Then the other issue, there is a lack of models. If you look at the Usher Syndrome Mouse, the Usher Syndrome Mouse is deaf, but the Usher Syndrome Mouse does not develop RP or doesn't develop the same type of RP as humans do. We haven't got a really good mouse model of the condition. Now, we do have bigger animal models, but bigger animals are harder to work with. This shows Usher Syndrome and it is deaf and it develops RP, but pigs are big animals. If you were testing therapies, you wouldn't get through the same number of tests as you would with a smaller animal that has a smaller life span. I think that's one of the key things is to get an animal that is small enough to work with, but it shows a good example of Usher Syndrome. So, the gene specific therapies is the Ush Stat. There is phase 1 and phase 2 trials. Phase 1, you're probably aware of the phases. You've got three different phases. Phase one is very few patients. Usually only a couple of patients. They're the patients with the worst possible vision. Basically people go into Phase 1 trials realising, it is not going to help me, but it might help other people who have the same

condition as me. They go into it just out of kindness, pure kindness, because they're going to see will this make a difference to someone else down the line? So, the Type 1 trials are small trials. They are only in one or two centres. Once you get to Phrase 2 trials, you're getting more people involved. You're not actually doing any harm by introducing the medicine. So, once they have confirmed okay, Phase 1 has gone through then they go to Phase 2. Phase 2 is more people and more centres. The study was on a big protein. They couldn't use the AAV virus. We have a gene therapy. The gene is different and it only affects the eyes. We can put in a working copy of the gene back into the eyes of those patients. Professor Keegan who I work with who is the head of our department, he's about to put the gene back into the patient at the end of this month, I think, into his first patient with this condition. It really does make a difference. Some Irish patients have gone abroad to have this. It has made a massive difference to their sight. It seems to have stabilised their sight. It seems to have even improved their sight. So, it has been a really big game changer. So if we could do something like that for Usher, that would be marvellous. Ush Stat Study. Basically they had nine people in that study. The Ush Stat Study, they found no improvement. At the end of four years, there was no improvement. I don't think they had worsened from baseline, but they hadn't got better from baseline. They decided, we'll try a virus that helps you put this gene back into the cells of the eye. So AV virus, it doesn't upset the immune system as much as other viruses and that's a good thing. It has a lasting effect. It stays within the cells that it is put in for longer. So, they started the Ush Study but in a different virus and they split them into two and they put them in the AV viruses in the hope it would get into the cells and work. We the haven't got results and we are waiting on those ones. There are only twelve patients. These are small studies. The other one I wanted to talk about. They found that for Ush 2A patients. A lot of mutations are in an area called Exon 13, if you have a change in that area, it will stop the gene from making the protein and then it makes a tiny protein that the body gets rid of. They can actually put a little plaster on the Exon 13 and skip it and read the whole gene and make a protein. It is really clever what they can do and it seemed to be doing a good job. Patients' vision improved. They stabilised their vision. The retina was more sensitive. The structure seemed to be improved. So although it is only 20 subjects, it seemed to be doing quite well. The only problem with this is when they

went to Phase 3, the company sold to another company and hasn't started the Phase Trials yet. So we are still waiting to see if that's going to happen. The gene independence study. This is interesting. These are tablets that you give to people. Basically, they're for people with functioning. With RP, your photoreceptors get damaged first and your codes get damaged afterwards. Basically, you can give this in a tablet form. It will up-regulate a molecule. It is a natural antioxidant. It actually stops free radicals and it is the free radicals which damage the cells of the eye. If you can increase natural antioxidants within the eye that's a good thing. We're looking forward to the results of those studies, but we have no results on them as yet. The other one is an oxen. Inside the photoreceptors you have molecules which absorb the light. You give that molecule in a virus into the eye. They're trying to resensitise the retina. There are no results on that study. They're due next year. Then you have another injection of **EA-2353**. This basically is trying to get stem cells which are present in the retina to be made into photoreceptors. So this sounds pretty cool as well, but there are only 14 patients and we don't have any results yet. Then the other one that I thought was really interesting was J Cells. You can inject them into the eye. They seem to stop photoreceptor death and seem to improve the function of working photoreceptors. They're happy with the results. 39% had an increase in vision and they're going to go to Phase 3 trials for that. The Photo Switch. The Photo Switch is a way of bypassing the photoreceptors and getting the molecule into the RPE where it can take things from there. It is a light sensitive molecule. Sorry, I've gone backwards. Okay, what can you buy over the counter and do they work? Vitamin A is something that has been considered that will reduce or slow down the retinitis pigmentosa. So this Vitamin A is easier absorbed. You can run into problems, if you take too much Vitamin A, it is toxic to your liver. So, our body doesn't store Vitamin A. It is something that you need to take in your diet. You just have to be careful that you don't take too much as it will be toxic to your diet, but out of the studies, we couldn't see any benefit for taking Vitamin A. Then the other one is lutein. It is a naturally occurring antioxidant. Some in the study said it was good. Omega 3. They are an ingredient of the outer segment with the disks. One study showed benefit. These are small studies. They're to the big groups and we need more studies on this. Then there is an interesting one on blueberry extracts. Blueberries are antioxidants. There was something that said it you took

blueberries, it would be with good for your eyes, but there are no studies done on that. Hold on. I'm messing up here. I'm going to run through what is happening with rare disease in Ireland. Because Usher Syndrome is a rare disease, you're part of the rare disease community. Your group, Usher Syndrome Ireland is one of a number of support groups that support people with rare disease in Ireland. Over-arching the rare disease support group, your own support group, is a group called Rare Disease Ireland. It is like an umbrella group for the individual support groups and they advocate the Government and they advocate for people with Usher Syndrome and all the other rare conditions to the government and try and get them to do things to improve life for people with rare disease. There is due to be a new rare disease plan. That's due to be produced over the next little while. We only recently had some lateral research ethic committees. You might who cares about Ethics Committees? Ethics Committees slow down research. If you want to get a clinical trial done, you'll have to go to each hospital board and it takes time. So, they set-up these national research Ethics Committees. One for medical implants and the other for investigational products. Like medicines. So there are two for medicines. The decisions of that will be national. So that is really important. That's going to speed research up really quickly. The other thing is that there is a rare disease clinical trial network. I don't know if any of you are aware of that. Trial networks try to get clinical trials to come to Ireland. I don't know if you know Rachel Crowley. She works in Vincent's Hospital. You could get in touch with this group and say this clinical trial is occurring in America, can we bring this trial to Ireland? She'll look for PIs and get them to be interested in that trial and bring it to Ireland. So, it is a way of making sure that these clinical trials are brought to Ireland. She finds the principal investigators to run the trials. She is also involved in a thing called RD Cat, trying to bring research into the hospitals. You have a rare disease... Let me just find... (Pause) So you have a rare disease research co-ordinator going into the five different main hospitals that make up the European Reference Network. The job of them is to seek out research opportunities and make sure that patients are available for research so they are consented. There are lots of things happening. For a rare disease register, we're going to end up having a register that is electronic based. Once the electronic records come in, we will have an author code. Every rare disease will have an author code. You'll get the full numbers of people in the country with a rare

disease because every single one of us will have an electronic health record. The European Reference Networks, these are networks of expertise. These got set-up for rare disease especially. These are all the experts for the rare disease. We're the lead centre. Basically it will be developing clinical pathway, increasing innovation and knowledge and helping with training and making sure that we are aware of what is happening in Europe and that we're all together in this. So, driving things forward together as a European country. These are all the ones in the yellow are the different networks. The ones in white are ones where we don't have a network just yet. So we have 18 of the 24 different networks that are set-up in Ireland with collaborations. Those clinicians and groups are collaborating very regularly with people in Europe. If you have a rare disease in Ireland and you can't get diagnosed, you can go to your network and they will give your case to Europe and all the European experts will look at your case. So you won't have to travel. So before what used to happen is you'd have to travel to all different experts, to go to different people to see can you diagnose me? So, what we've been tasked with is to provide a register. Although we have the Target 5,000 register, we're creating our own register which will marry up with the European register. The rare eye disease register, we're in the process of producing and we're using the terms and the author codes for that and the inherited retinal disease register will be part of that. National care pathways are very important. Usher Syndrome does not have a national care pathway. Care pathways where an idea of the national rare disease office. They produced 30 care pathways. A care pathway is a way... I've done something serious now. Help! Care pathway is a way of bringing a clinical pathway and patients and clinicians together. And together you're deciding what care is needed for people with this condition? Who provides the care? Do I want your care to be provided locally? Is it better that it is provided locally or in a hospital and where does it happen? So, there are 30 care pathways that are published. There is one for RP which I'll just show you. (Pause) This is the one for RP. It tells you what happens at diagnosis. How should it happen. Who should tell you your diagnosis. What happens. Who do you go to? How do you get diagnosed. That's the first thing. Then what happens in the hospital. Then what happens in the community and then what happens in primary care? So all of those things and these are HSE endorsed. You want your pathway to be HSE endorsed. The chief clinical

officer has signed off on all of these. So if you get your pathway, he said that this is the way that your condition should be managed in the country. So that's hopefully fund will be provided then as part of that. I don't know if I've time to go to my case study.

KATIE: Yes.

JACQUELINE: This is a little boy, Sean. He's in pink. He was born and he congenital bilateral sensory deafness. At ten in the Children's Hospital, they started doing panel testing for all people who had congenital deafness. They've only been doing it for the past years. They look at the 100 genes which cause congenital deafness. The Usher genes are within that panel. He got his results back. The gene that he had - he had two mistakes on his genes, but that gene could cause either congenital deafness or congenital deafness and retinitis pigmentosa. His mum and dad were very upset. They weren't sure. He's got deafness now, but is he going to run into problems with his vision? So, we looked at the gene alterations and we organised for him to be seen by the eye consultant. It is a traumatic time for parents. Do they have full knowledge of the genes they're being tested? Do they know this is something that might come down the line. "Your son has deafness, but in time, he might end up going deaf as well." There will be a period of time that he has sight and it will be very difficult for those parents to see all the things that he's able to see and realise that one day he's not going to see them. So, you know, I wanted to bring that up because it was a difficult one when we saw the people in clinic. One of the changes they hadn't been seen before. We were confident it wasn't going to reduce a protein and the other gene, had been seen in people with deafness and also in people with Ushers. We were not sure is this going to cause Usher Syndrome or not. For people with Usher Syndrome, I will go back to this one. If you have a child with Usher Syndrome. This is a type of picture. There is a one in four chance of another child in another pregnancy having Usher Syndrome as well. Your other children who don't have Usher Syndrome, they have a two in three chance of carrying the gene for Usher Syndrome because both parents carry one gene. They have one gene that works and one that doesn't work. The other children will have a two in W3C. If they marry somebody from the general population who has got no history of Usher Syndrome, the chance of them having one of the genes for Usher Syndrome is one in 60. What we're saying is that there is a much, much less than 1% chance that their children will have Usher Syndrome.

So very, very small chances, okay. The same for somebody who has got Usher Syndrome themselves. Because you have two genes neither of which work, you are always going to pass one gene with Usher Syndrome on to your children so they will all carry it, but your partner, if they're from the general population and don't have any Usher Syndrome in the family, the chance of them being a carrier is one in 60 which means there is a less than one percent chance that any of your children will have Usher Syndrome. Saying that differently means there is a greater than 99% chance that your children will not have Usher Syndrome. The chance of anything going wrong in any pregnancy is one in 40 or 2.5%, you need to balance these figures with what happens generally. They're considered very low risks. A slide on the supports that are available. You've got your wonderful Usher Syndrome Ireland and they're part of #RARE disease Ireland. You've got the Rare Disease Office. If you're worried about care or stuck and don't know who to turn to, you can ask the Rare Disease Office. They have an information line. They get involved in making sure that Ireland gets all the grants that it should get so we can support our rare disease patients. You've got Fighting Blindness. You've the Guide Dogs for the Blind. You have therapeutic counselling and Vision Ireland offer therapeutic counselling for people who are losing their vision. It can be a very difficult time. And Fighting Blindness have started therapeutic counselling. It is where they offer ten free sessions. If that was something you needed, it is available. There is a good podcast called The Blind Guys Chat. It is quite funny. It is quite good. You can contact them and find out more about them. There are quite a number of those podcasts and Vision Sports Ireland. There are Sports Inclusion Officers around the country who are trying to get you involved in sport. If you want to do sport, get in touch and say, "I want to do it. I want to try." I'd like to thank the team at the Mater Hospital. A wonderful team at the Mater. I want to thank the teams at Temple Street and the research lab in Trinity which help us when we're stuck and at the moment trying to help us with patients that we couldn't find a gene change for. I'd like to thank you, our patients because we learn so much from meeting with you, from talking with you. You give us feedback and that really helps. I know that at one stage people found it difficult to get through the hospital in the Mater Hospital because very hard to find the units. We took that on board and we have nice, big placards telling you where to go. There are only three areas. They don't complicate

it. There are big signs so it is so much better to get through the Mater Hospital which is a maze at the best of times so we do take on board what you tell us and improve things. Thank you very much. (APPLAUSE)

DR CONAN DONNELLY PRESENTATION:

Good morning, everyone. Thank you so much for inviting us to this meeting and we're really pleased to be here to talk about a project that Usher Syndrome Ireland have asked us to undertake about the development of improving data solutions for the Usher community in Ireland and beyond. My name is Conan Donnelly. I'm a director of Connect Research along with my colleague, Jackie Boylan who is a psychologist and we undertake research to support patient organisations and other clinical research and particularly rare disease. I have worked - my expertise is not in Usher Syndrome by any means, I work in disease registries. I was really interested in Jacqueline's talk earlier. I'm really pleased to hear about the progress that's being made in Ireland. I worked in the National Cancer Registry in 2020 and the progress that's been made with the healthcare records, and health identifiers is amazing. It is going to make a huge difference. I believe that's going to really transform the healthcare in Ireland. One interesting thing about cancer registries, if I go on to the National Cancer website, you can find out how many people were diagnosed with bowel cancer last year and Stage 1 and Stage 3. We cannot say how people live with Usher Syndrome. This is the case in most countries. There are a handful of exceptions which are ahead of us. England have developed an excellent disease registry. It is early days. I'm getting ahead of myself. Exploring the data needs of the Usher community in Ireland and beyond. It is a scoping project. Looking at the options and potential solutions including looking at the existing data landscape. Jackie has given a detailed presentation on that already. We're talking about data needs and thinking about the data needs and solutions. What is a patient registry? They have been defined by the European Medicines Agency as an organise system that uses organisational methods to collect data and follow it over time. That is a fairly dry definition. It is a useful definition. Particularly the uniform data so that things can be measured consistently. I want to take a step back from that and look at what you need to build a registry. You need a community of patients, families and clinicians and researchers, creating a culture of collaboration and engagement among a broad range of stakeholders, to create fertile ground for research. That's really what the benefit of a registry

more than anything. It is not necessarily just the data, it is creating that culture and collaboration. So, patient organisations and when communities decide to create registries, there are many reasons to do it and there are many values in creating registries. In rare disease this has become a Holy Grail for patient organisations. First and foremost a contact registry, knowing who the patients are, where the patients are and a little bit more information about them so you can define the population and understand the challenges as a community or nationally, we face in addressing their needs. Finding people to join clinical trials. That's the minimum of what would be expected, but it is actually really, really important. Another area is prevention of the siloing of data. What do I mean by the siloing of data? Data collected for a specific purpose, let's say a clinical trial or study, but is not available to other researchers and users. One of the things the registry can do is promote the secondary use of data. And then there is a need to use that data again and again and again when the registry is well set-up then that data can be used not just for one study, but for lots of studies. I suppose that's what patients want when it comes to their data. They want their data used and used well. Looking at how patients want that data to be used. That's really important in rare disease. It is not just the disease that's rare, the data is rare and the data is well evaluated. Also to promote a culture of collaboration. I've already spoken to that and create the research infrastructure that is under-pinned by community control and governance of data. What does that mean? That means that the stakeholders that are interested in making this data work as hard as possible in the interests of the patients. Another important use of it and this is at the later stages of clinical data development, is this drug effective? Registries are very valuable for that because they collect data in the real world. They collect data from a range of sources, not just from the clinic, but from the patients and to demonstrate if a country or health system invests in this drug, it will be cost effective and it will be beneficial to the community. Informing study design. Natural history studies are really, really important in informing design for clinical studies. How many people do we need to recruit in a clinical trial? How should we define the population in the clinical trial? This information is particularly important and most often and best done through natural history studies. Registries recognise a potential source of that information. Sometimes going beyond that, registries and natural history studies are sometimes sought as an

option to avoid placebo controls and replace them with a natural history control and some people see registries as a source of that, but there are only a few examples of where this has been successful. Informing guidelines and clinical guidelines. What's happening on the ground. We heard about the standards of care that have been developed in Ireland. The evaluation and monitoring of that care and is it working well? Is it generally happening on the ground? Support audits and things like that. So patients are getting a good quality of care and registries are often used in that respect as well. Finally, and often and this is a need in industry, for post-authorisation, when a drug has been approved for safety and long-term. Does it work in the real world? Is it as safe as it was in the clinical trial? Because the real world is different. It is not an experimental environment. So, those are some of the reasons why registries are collected. There are a myriad of reasons. You can see there are all sorts of reasons why a community might establish them. We want to understand what are the priorities of this community? The design of the registry and the model of the registry can be very different in different scenarios. I really want to focus on this. There are some real challenges in establishing registries and the vast majority of registries do fail. That's a reality. There was an examination of registries in Ireland a few years and it was found something like 200 registries, many of which had closed down. There are many challenges that I'm going to talk through. Building and retaining community buy-in. That's a must. When I say community buy-in, I mean the broadest sense of community, all the different stakeholders who need to come together. Working internationally can be a real challenge. It is very important especially ultra rare diseases where there are maybe not enough patients in one country to collect enough data to make it possible to answer certain research questions. Working internationally comes with challenges. There are different regulations and different ways of doing things in different places. There are also different cultures as well. So different expectations from different communities. Registries need to be responsive to be useful. So, often it happens, registries have been established and don't fulfil their research objectives and the data ends up not being shared or not being shared quickly enough. So having a really, really clear understanding of how a registry or a data collection works so it will actually foster research and not get in the way of research. Building and maintaining trust and confidence across all the spectrum of stakeholders. Particularly the

professional side so they're willing and happy to contribute and support is something that is always a challenge and this is the one of the reasons we have siloing of data because there are many reasons why organisations, for commercial reasons, don't want to share data. There are many reasons why academics may not want to share data as well. This is a well-established challenge. We are improving on that and we are hearing of much progress being made in terms of having a more positive attitude towards sharing data and being more willing to share data and setting up systems to share data, but registries need to be established in a way that creates that model. Maintaining independence and this is when it comes to working with the commercial sector. The sustainability of registries with the support of industry to fund that and of course that comes with challenges of ensuring there is a trust that the registry still has its own values and interests at heart and is not leaning towards one particular company or another. So having a real strong governance model is a solution in that. And then competition. When we think of registries, think of what registries try to do. We try to bring people together and try to bring data together so that we have all our data in place, but actually what happens sometimes is there is lots of different registries. You have a registry for one specific condition, but then another registry for a group of conditions including that condition. What can happen here is you end up having the same silo of data, but with a different name. They're all called registries. This is something we're exploring with the community to understand how best to address these data needs to avoid those potential pitfalls and problems in the future. When you can see the things that registry can do. What are the most important goals to target so they are achievable and realistic? So, I haven't mentioned central issue there and that's one of thought. For any data collection system to be useful, they have to reflect what the true experience is of patients. They are designed to put a mirror to a community to a rare disease and say and provide statistic information for that condition. For a registry to do that well, it needs to have high-quality data and needs to be up-to-date and needs to be rich in detail. Now, we have two images here. One on the left which is a pixelated of the Mona Lisa. Without complete detail, without a complete picture you have this pixelated image. This may be reflected statistical outputs because you haven't got enough numbers. Your understanding of the statistics is actually weaker than you would like. It may reflect missing groups of

people in the population or missing parts of the patient journey. So, the rich picture featuring all the elements of the Mona Lisa is the one we want to strive for on the right-hand side. Many patient registries have been established which have been more like the picture on the left rather than the picture on the right. So, I think striving for something that builds that picture up drew having a high-quality data system to have a really good understanding of the disease. Related to that is having a registry system that allows you to collect the data in a way that reflects the need. So, if you want support in a natural history study that's going to contribute to a clinical trial. A registry isn't the only solution that we are looking at. There are other things that we need to consider. One is seeking access or encouraging wider access to existing databases. Linking and combining data. So, for example, there is some rich and valuable studies around, but if they're not linking together, we don't have a complete picture without having all the data. With Usher Syndrome, it is often the case that we've got data on the RP, but maybe not information in the same database on the hearing. As we find out more about Usher Syndrome, we find out maybe that there are other aspects of Usher Syndrome that we need to capture information on as well. So, connections of the data at the individual level, the individual patient level is very, very important and there are some really interesting approaches being taken to facilitate and foster that and this isn't necessarily just for research. It has got wider benefits. In unconnected health systems, patients who are transitioning from maybe a childcare into adult care can actually have the patient information combined and this is something that I'm particularly interested in developing resources in. Digital technology, this is something that's really interesting. Your wristwatch or information can help understand the impact of the disease in real-time. Think of your Fit Bits and devices like that, but devices which have been clinically validated. Is this the right time to start to look at things like that? These are questions that we will be exploring. We want to hear from you. We want to hear what your thoughts are and what is important to you in the Usher community. We're providing some information. We're going to share some questionnaires with you and we're asking you to provide some feedback and this will be anonymous and we're going to summarize the responses in the work we're doing for Usher Syndrome Ireland. We're really grateful for your insights. No research is possible without the patient community and the carers. So, I think it is always

important to ask what it is that you want from the research. So, I think that's my presentation. Thank you very much for your time. Thank you very much again to Usher Syndrome Ireland for having me. (APPLAUSE)

DR SOLOMON KAMAL-UDDIN PRESENTATION:

Hi everyone. To start from the beginning. My name is Solomon. It is my pleasure to have this talk. Thank you to Carol and Usher Syndrome Ireland. I'm sorry I couldn't be there. I'm hoping to spend the next 15 minutes talking at an overview level of what it is like from the medical point of view in terms of expectations patients should have when they get diagnosis of a rare disease, in this case, Usher Syndrome and what their hopes of the medical system afterwards. You guys are experts in your own right, but at the same time it is always good to be armed with more knowledge if that's okay. A small bit about myself. The rare disease journey if you have come across the wider framework of rare diseases, it is something that's very well defined. All the members of the medical team and healthcare professionals that can get involved and any questions I'm happy to take. So myself, I'm just a paediatric consultant. I've been a paediatric consultant for nine years. Practised in and around London. I moved out in the past year. My training is actually as an Emergency Medicine specialist. I used to run an A&E department as well as do general paediatrics. What that means whether I like it or not, I have the training and been privileged to be part of diagnosing rare disease and genetic conditions, working with families, but in terms in working in A&E where some of you may also be aware. A lot of frustrations that can happen when you have a chronic disease, sometimes a chronic disease may have subtle problems or symptoms and may not be diagnosed yet. As well as the GP, A&E is often the first port of call for many of these families. I have experience as a professional so the different aspects that can happen in that rare disease journey before diagnosis. Also in my time as a paediatric consultant, I have helped design and recruit clinical trials and worked with international organisations. I helped launch drugs in rare disease. We know as a medical profession and as a drug industry where there are some big, big gaps in certain patients and it is very satisfying to be able to bring treatments out the door to where they're needed. I have supported work under the emergency framework. This may or may not be something you and your members have heard, but the best bit of black and White Paper there is in the UK in terms of government support for rare disease and back in 2021, there was an up-to-date

document divided into UK, Northern Ireland, England and Wales and Scotland and it did include aspects of Ireland about how rare disease should be addressed. So our story is everything seems fine and during the pregnancy, the scan was more specific. We came to Great Ormond Street Hospital and the youngest daughter would be very sick. What was interesting we were recruited into rare disease trials and genetic trials. I've involved in the 1,000 gene project. We're privileged to be part of that journey as well. The rare disease journey which Usher Syndrome is. The medical story is one which needs more education for students and junior doctors and for healthcare professionals. The hope is that with education some of the frustrations around being a patient with a rare disease may be addressed. Yes, the idea is that rare diseases are rare, but collectively one in seven people in the whole world suffer from a rare disease and all together rare diseases have particular characteristics in the fact that they're under-served. There is not much education about them. They don't have many treatments for them. Patients are particularly vulnerable because of the lack of medical support and medical knowledge and they tend to be under-diagnosed and they're spread across different specialities. On average there is a statistic that says it takes a 7 to 15 year journey to get a diagnosis. From the moment when someone might have lots of subtle problems and sitting in front of a doctor saying this is what you've got might take up to 15 years. It depends on, a lot depends on. In that 7 to 15 years, you see multiple medical teams. You are talking to your GP and A&E and the neurologist. Many aren't able to pin down what you might have. Every time you see a different medical specialist, they don't know what you're talking about. You have to start the story from scratch and the same again with your friends and family, you have to tell them over and over again about 20 problems which don't seem right. Not many people have the same set of problems. People can end up being emotionally isolated and physically as well. In some cases, it is hundreds of miles before there is another professional with the same set of problems. The symptoms, as in Usher Syndrome, means that you end up feeling isolated because there are so many additional communication hurdles. And challenges with daily functioning. The world isn't built for people who are a bit more interesting and a bit different. There are challenges every time you want to just get out the door and do something. What's the flip-side after the diagnosis? It is probably not much different.

We see multiple medical teams several times and you see different doctors who seem to be responsible for your care or not responsible or different health professionals. There is a lack of knowledge about your symptoms among doctors you might see and when you tell members of the public who may not be aware of Usher Syndrome, they're ignorant about what it is and you have to explain what it is over and over again. That physical and emotional isolation, but it might be a bit better, but the challenges of a rare disease still exist. The one big difference that happens is you've got an added frustration where now you have a diagnosis, there is a lack of available treatments. Okay, I've got it now. So, what's the cure? You're told actually there is not much of a cure. That's some additional considerations, but again I'm hoping that I'm not preaching to the converted here. So, who is this team that are responsible for helping support you in your medical problems? There is the hospital specialist. I actually know the hospital Specialist who is responsible. He is a wonderful guy, but very, very busy. Responsible for delivering a diagnosis and resources. This is the perfect situation, resources to offer a follow-up appointment. Having that news broken to you, sometimes after a long wait, you take two or three sentences in and after that is a bit of a blur. The ideal thing is to be able to speak to someone two or three months down the line, just in terms of helping someone understand what it is they're facing moving forward. Ideally the hospital specialist can offer a follow-up and know about the disease and the science of it in terms of the prognosis, in terms of treatment options, specialist treatment options and that long-term follow-up might be six months, six months, sometimes yearly is the best way to co-ordinate discussions around how is your disease going? What's going to happen with your disease? Oh, this treatment is coming along. This may or may not make a difference. There is the co-ordination where they have the best understanding of the disease and symptoms. The hospital may or may not have a set-up to be able to talk about them. Not only do you have this disease, but you've got your eye problems. You've got your hearing problems. You've got moving and mobility problems and having that knowledge means they can talk about getting those hospital services in and those hospital services in and some of the follow up appointments is also not just giving you information, but making sure the co-ordination of the whole package is happening. If there are special problems related to the disease, some rare diseases for instance you've

got bone problems which might need operations or problems with the ear and the hospital lead can say bring in ENT earlier and I can write that letter easily to my colleague in the same hospital and I can move that forward. There should be a repository with all the information. Unfortunately, this is not always the case, but the hope is it is on paper with all the information around your disease journey. Primary care and the GP. A lot of rare diseases where people need support. It actually comes down to their GP in terms of making sure things are in place on a monthly basis to help you move forward through that. So if physiotherapy is required. Then the GP is sometimes the one who is best placed to co-ordinate the services and if that care drops over a day or a week or a couple of weeks the then the GP is best placed to facilitate that forward. If there are repeat medical prescriptions, it is the GP's responsibility to main sure prescriptions are maintained. If there are queries relating to the disease such as ear infections then they can be the first port of call. They should have knowledge of the disease and that is a challenge. GPs have to know everything about everything and having a rare disease on their plate and knowing the ins and outs of it when they maybe see one of these patients every three or four is a challenge, but they should have a working knowledge of the disease and have conversations around what is happening to you? Talk with the Specialist as to when things get bad. They should be taking that information and forwarding it on to the hospital specialist, all that communication and knowledge about what is happening should be going to the hospital specialist. You have got two different roles. One is more the community, functioning and the other is to do with the disease overall. You have heard about genetic counselling. That's for the patient and patient's family. If the hospital is lucky enough to have a specialist nurse, then all the conversations about the follow-up appointments, who do I go to see X about Y, you've forgotten my medicines, a lot this can be taken up with the Specialist nurse. It is a powerful, but rare resource to have. A&E is a challenge. There will be emergencies in chronic disease. There will be that conversation where you have to tell them from the beginning about your disease. The eye consultant and dietician and you've got the audiologist. I want to emphasise the mental health aspect. Everybody who gets diagnosed with chronic disease should have access to some kind of mental health. So, I hope that gives the structure. I will finish on my last slide. People with rare

disease who have had it for a while sometimes get sick and tired of having to be the one to carry the luggage and are told you need to carry more. This slide is about how proactive you want to be, but if you want to take a stronger role then absolutely. Keep a healthcare folder with your records and information. If you come to a doctor who doesn't know what's going wrong or a physiotherapist or optician, you've got a one page print-out. That can be a huge help. Get involved with organisations, Usher Syndrome of Ireland. It doesn't have to be disease specific. There are other rare disease organisations like Beacon are happy to take on board and have conversations. Be proactive in seeking mental health support and have the conversations with your clinician about clinical trials because that's another way you can get involved. That's all I had to say. I hope that was useful and I'll hand-back. (APPLAUSE)

KIDIST PUIG-LYNCH:

Hi. My name is Kidist. I hope you can hear me. I'd like to thank Carol and her team for letting me have this excellent opportunity to speak. My name is Kidist. I'm 14 years old. I was born in Ethiopia. I live in Wicklow. My dad is Irish and my mum is Catalan. I speak English with my dad and Catalan with my mum. I learn Irish in school and I learnt Irish sign language when I was younger. I like reading, horse riding and playing basketball. I found out I had Usher Syndrome when I was eight or nine. I have Usher Syndrome Type 1B. I don't remember anything about my diagnosis. I just remember I couldn't see anything in the dark. I don't remember not having it. It has been such a long time since I got it. I got my implant in Temple Street Hospital and my second implant in Barcelona. Usher Syndrome is a big part of me, but it does not define me. My parents have always pushed me to be independent and have high expectations. I don't think about it that much and I don't get constantly depressed about it. It is an inconvenience and I do get frustrated sometimes, but for example when I go on an outing with my class, and maybe parts of it are dark and I need assistance, I find I'm holding people back. I find my deafness is an inconvenience. My visibility is only noticeable at night-time. My lack of balance was more annoying when I was younger. When I tried to do handstands and cartwheels and I couldn't, but now I'm older everyone has calmed down a lot more. My family are very supportive of my disabilities and they're always willing to help me. It is hard without my family especially at night-time. I need time to be aware of new surroundings. Now that I'm getting older, everyone likes

going out more at night to get a sense of freedom and this is more difficult for me and it can cause a lot of hassle as I have to say no to a lot of stuff. Family occasions are tricky. We have big family dinners. Everyone is talking at the same time and there are multiple conversations going on at the same time. I feel over-whelmed and frustrated. It helps when I sit with my back to the wall so I'm not getting so much background noise. I also have to ask people to explain what is going on. It is better if people are divided up into small tables. I started secondary school a year ago. Before I started first year, myself and my parents had a meeting with the school with the Visiting Teachers who ensure it is a safe and accessible space for me. In those meetings I got to walk around school and see what it is like which was great for me. A few weeks into first year when I was more comfortable in this environment, I sent an e-mail to all my teachers listing the things I needed help with, what was going very well, etcetera. Among the examples. In one of my first classes in first year, my home economics teacher told me she was worried about when we were cooking that I was going to, apparently somehow disconnect the oven. It was very hard not to laugh in her face! It was very funny. So, I sent her an e-mail to remind her that's not going to happen. This e-mail contains so much. I'm so much more than a new deafblind girl. I ended up doing very well in school, doing top of my class and winning awards. I ended up getting Student of the Month in September and I also got Student of the Year for Spanish in first year. I think Visiting Teachers are very important. A good visiting teacher is not just there because it is their job, but they want to see you grow. They advocate in school and make sure you have support that you need to learn. Visiting Teachers are very encouraging, on the topic of visiting teacher, there is my visiting teacher, Jan in the audience. I'm so lucky to have her. (APPLAUSE) I enjoy a lot of sport and activities such as basketball and horse riding. A big problem I have in contact sports is the ball hitting me. I don't see the ball coming, but I don't let that stop me. Having Usher Syndrome, you have live life to the fullest. This didn't come easily. I try to push myself. I have learned that people who are willing to understand your needs. If I go somewhere that's going to be in the dark, even though they know about my Usher Syndrome, I remind them so it is fresh in their mind and they are fully away. Usher camp is organised by Usher Kids UK and Ava's Voice. This year it took place in Ghyll Head in Lake Windermere. It was my second year attending the camp. Usher camp is amazing. You get

to meet so many people with Usher Syndrome. When I was there for the first time, I was nervous because I was away from home. I got over my nervousness. A group of people with a rare condition having fun and bonding and forming friendships. The whole week was fun doing archery, caving and so much more. When I was asked if I'd come back next year, without pausing for thought, I said yes. I'm so happy to be part of this welcoming, positive community. Ever since I was little, I wanted to be a teacher. I love the idea of passing on my passion. When I grow older, this may change. I want to be inspiring and a role model for people who look up to and for them to think, "Well, if she can do this then I can." I always loved school and I have had some amazing teachers which make class fun. Three things I wish people knew about Usher Syndrome. Things that maybe easy for you may be very difficult for me. Things that can be easy for you to see, can be very difficult for me to see. When I tell people I have Usher Syndrome, I don't want them to call me Angel Box. I want to get out of that box. It is really hard to deal with large groups of people. There is just too much going on. Keep it small. Something I do when there is a large group that is very loud like for example in school, in my class, I take breaks. I am allowed to step out of the class for a few minutes and gather my thoughts and just calm down a little bit and then I just go back into class and that is a very effective way to help me. Three things I wish I'd known early. If you're given the opportunity to do something, just do it because you may not be able to do it in the future. I love trick and treating and I didn't know I wouldn't be able to do it in the future. I'd advise for you guys to make the most of it. Be good people. Good people attract good people. People are willing to help you. I try and give as much as I can. I try to be a good daughter. A good sister. A good cousin. A good student. Then when I need help, people think, "Oh she is a really nice person and she needs help now. She is such a good person. She is so lovely. She is so kind. She is so nice." I really want to make her life a little bit easier and living life to the fullest, don't worry too much about the future. Don't worry about the future. Just throw yourself in to new experiences because from my experience it has helped me a lot mentally as well. It makes me stronger mentally and when I get something I don't like, I can handle it a lot better. For the parents I know it is difficult your child getting diagnosed with a life changing condition. They're the most important thing to you and all you want to do is protect them, but the only real way to protect them is not to wrap them this bubble

wrap. Just make them go to new experiences like Usher Camp. Throw them out into the world so they can experience it and not hold themselves back because when they're younger they didn't have much experience. Having Usher Syndrome may not be the best thing in the world, but I feel positive, different and unique having such a rare condition. Going to Usher camp and being part of the Usher community makes me feel so welcome and supported. I would like to thank Axon for sponsoring my presentation. Thank you for your time. I'd like to show the media of Usher camp. I'm happy to take any questions. (APPLAUSE)

'ARE YOU USHER AWARE?' LAUNCH

CAROL:

Thank you everybody. It has been a very good morning. We have run over time. We won't rush this important part. It is another part of the day which has been very important to Deborah, myself and the Board over the past few months. Can everybody hear me? Can we please keep the noise levels down because some people need to hear. Okay. Thank you. We were very lucky to receive a grant that is known as the Rare is grant from a pharmaceutical company called Amgen. A healthcare pack will be sent to all healthcare professionals in Ireland. North and South. In order for the Usher community to have a more positive experience when attending medical appointments. So, I would like to welcome John Barbour who is the director of medical affairs of Amgen to formally launch our awareness pack and I just want to describe the contents of the awareness pack. So, doctors and audiologists will receive a box in the post and inside the box we will have simulation glasses so that they can understand what tunnel vision is like. They will also have earplugs to wear with the glasses so as to get an understanding of the challenges of hearing and sight impairment statement. We will have other information, patient leaflet books, books for healthcare professionals that goes into the more medical terminology of Usher Syndrome. We have some labels. We really hope to spread the word. If you haven't sent us - at the back of the room there are two lovely girls collecting the names of your doctors, audiologists and eye consultants. Just give us the name and the location of these people and we will happily send these packs. This is huge thanks to Amgen to make our lives much better when attending medical appointments. We also have to thank Amgen and I'm proud to announce we recently were successful in receiving

another award and we will be using this for one-to-one coaching. This is not counselling. This is coaching. It will help you to try and set goals in your life and work towards achieving them. So, we will be announcing more information about this coaching service that we hope to introduce very soon. Thank you, Amgen. Please welcome Jon Barbour.

JON: Thank you for the warm welcome. We're delighted with the initiatives that you're going. Kidist, an inspirational presentation. Keep up the good work. (APPLAUSE) Jon Barbour is my name. I work with the Medical Affairs Team for Amgen Ireland in Dublin. I'm connecting to you through the hashtag RAREis programme. It was launched in 2018 to elevate the voices and experiences of individuals across the world and especially so for us in Ireland. In 2022, we launched the hashtag RAREis global advocate programme. This pack was launched today. I had a look at it and it will be a benefit to families and patients with Usher Syndrome. You can imagine now healthcare professionals throughout the nation being better informed and therefore hopefully improving that patient experience when you go to your clinic. That's really important. Back in February we invited Carol to share your perception and insights. I was there. What does it mean to have Usher Syndrome? What does it mean to the community? It highlights to me personally the importance of community and cultivating this community and what comes out of it and the importance of research. Thank you again for joining us that day. Amgen are proud to sponsor activities like this. To sponsor support groups like Usher Syndrome Ireland. We're really proud to do that. We're proud to support this really important initiative which has been introduced already. I'm not going to hold you back from lunch any further. I'd like to congratulate you in the realisation of such an important initiative. If you want to learn more about RAREis programme, go online: Www.rareiscommunity.com. Thank you for inviting me today. I learned a lot and I appreciate you sharing your time with me. Thank you. (APPLAUSE)

CAROL: Now we will bring the morning session to a close. I'd say our interpreters are really and truly ready for a rest. We have to say communications, accessibility, is absolutely really appreciated and it was funded by the Anne Sullivan Foundation. Without the Anne Sullivan Foundation, we would have done our best, but the Anne Sullivan Foundation did a lot with the help of Marea, our stenographer and our interpreters, all of you, thank you so much, and last, but not least, Michael down the very back of the room. Thank you so much, Michael, for just

making all the technology work. So, I would like you all to now get ready. We need to be outside and starting to walk towards our afternoon venue. We will walk together. So if you can quickly go outside and into the square. We'll walk over as a group to our afternoon venue for lunch and other activities. So, thank you.