

**(Mark Dunning)** So, can I ask Emma, and Laura, and Isabelle and Brendan to come on up here? We are going to do a combined family and researcher panel.

And I am going to moderate this panel. And I am going to ask some questions. But if we have time at the end, I will open it up for questions and answers from the audience as well. Okay, so I am going to ask everybody to introduce themselves briefly. Brendan, we have you go last for introduction. As I know, you have a little bit more you want to say, okay? So, I will start with Emma, do you want to introduce yourself?

**(Emma Boswell)** Hello everyone, my name is Emma Boswell. I am from the UK. I work for a charity called SENSE. I am also the chair of the DBI Usher Network, and I have also set up a deaf cancer support group. I have Usher I myself, and I am married with children, and I am not sure, if you need me to say anything else? Thank you!

**(Mark Dunning)** That's perfect. Isabelle?

**(Isabelle Audo)** I am Isabelle Audo. I am a clinician scientist, working in Paris at the Institute de la Vision and in l' Hôpital des Quinze-Vingts.

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**(Mark Dunning)** Are you married? Do you have kids?  
Where do you live?

**(Isabelle Audo)** I do, I have kids. I live in a suburb of Paris.

**(Mark Dunning)** Claes?

**(Claes Möller)** My name is still Claes Möller. And I am an ear and nose and throat specialist. And I specialized in audiology. I am trying also to be a vision doctor, but they don't like it. I have 3 kids and 2 grandkids.

**(Mark Dunning)** Laura?

**(Laura Bingenheimer)** Hello, my name is... can you hear me? Yeah, okay. Hello? May I use that? Okay, thank you! Hello? My name is Laura, I am from Munich, and I am doing a fundraiser for certain research projects. So, I am actually building a bridge between the researchers and donors. So, yeah, that is what I am doing. And I do have Usher myself, Usher 2A. Yeah, that is why I am here.

**(Mark Dunning)** And Brendan, did you wanna talk? You had a little bit more to say, correct? Or do you just want to introduce yourself?

**(Brendan Creemer)** Yeah, I will just introduce myself. Hello, everyone! My name is Brendan, I am 19 years old and I have Usher Type 1F. And have taken some action to spreading awareness and fundraising which I will be talking about soon.

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**(Mark Dunning)** One of the nice things about this panel, and the reason we asked for the researchers and some of the families to sit together is: I would like to ask you guys some questions. I really kind of want the audience to see that you guys are really not all that much different. I will ask Claes and Isabelle: If you can answer these questions on a more personal level, as well as related to what you see in your careers? I would be interested in hearing that. So, I start again with Emma. And Emma, my question to you is: What do you worry about on a daily basis?

**(Emma Boswell)** Well, I worry about not being able to communicate with my children because I have Usher and I am an Usher parent. I believe, that is one of the biggest challenges, I face every day. Oh, don't take me wrong, I try to be positive, but that is probably my biggest worry.

**(Mark Dunning)** Don't worry I will give you guys an opportunity to be positive. So, Brendan, what do you worry about on a given day?

**(Brendan Creemer)** Sorry, can you repeat that?

**(Mark Dunning)** What do you worry about on a normal day?

**(Brendan Creemer)** I actually try not to worry because I am confident that a cure will be coming out within my life time. I just try to about life every day as if I don't have Usher syndrome. Well, keeping an eye on the research, making sure that it is making progress, like looking out

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for the cure which will-I know will come some day. (applause)

**(Mark Dunning)** So, Laura?

**(Laura Bingenheimer)** I am totally with you!

**(Mark Dunning)** You don't worry about anything at any given day. There is nothing that worries you? You are just carefree?

**(Laura Bingenheimer)** I don't really worry because I think, it is not leading me anywhere. Focusing on the research is great, that it makes progress, but on the other side, I am just living my life, and doing the things that interest me a lot. Because I also have to say that I am not as restricted as some of you are. So, I can communicate and do everything like that. So, I am still very happy actually with that, yeah.

**(Mark Dunning)** Great, thank you! So, Claes?

**(Claes Möller)** I worry about that still a lot of people with Usher syndrome don't get the right treatment and rehabilitation. I worry about that it takes such a long time from research to a clinical practice. And I worry about that I have to retire. (laughing)

**(Mark Dunning)** You don't have to retire.

**(Isabelle Audo)** So, you mentioned on the personal basis.

**(Mark Dunning)** Yes.

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**(Isabelle Audo)** So, I try to not worry about anything, but I still do worry. I worry about not being able to do, what I was expected to do on a daily basis. And I have been worried about, you know, missing a diagnosis, or think something that I shouldn't say to a patient or to colleagues that will lead to other worrisome things.

**(Mark Dunning)** That is a good answer. So, my next question for you guys is: How do you feel, when you meet someone new who has Usher syndrome?

**(Brendan Creemer)** What?

**(Mark Dunning)** How do you feel, when you meet someone new who also has Usher syndrome?

**(Brendan Creemer)** Well, I try to relate to them in some ways. But for the most part, I try to motivate them to take action, to follow the research, to keep an eye out for the cure, spread awareness, like what I am doing.

**(Mark Dunning)** Thank you! Emma, do you want to answer that question?

**(Emma Boswell)** I was very excited actually to meet new people. Because we are so varied, and I like to see, what other people have achieved in their life, and that actually empowers me.

**(Laura Bingenheimer)** I don't know really know many people with Usher syndrome actually.

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**(Mark Dunning)** You do, now. *(laughing)*

**(Laura Bingenheimer)** I do. But when I meet them, I think it is the same thing as always, when you meet people: you have some things in common and some things you don't. So, that is just one more thing that you have in common.

**(Mark Dunning)** Excellent, now you guys deal with Usher syndrome all the time.

**(Claes Möller)** Yes, and when I meet a patient with Usher syndrome and we have cleared out the problems, then I try - because I usually have 3-hour sessions to start with - I usually try to also find all the strength and all the beauty in being a human being, and that people with Usher syndrome are as different as all my other patients. And that is so rewarding.

**(Isabelle Audo)** So, when I meet a new patient with Usher syndrome, I try also to see the positive things out of all the other things. You know, after the testimony of Julia, I really always keep in mind that maybe this patient has a history of bad news, good news or a deception and so on. So, I try to read with the patient this story. And also to give an example of people who despite everything achieve great things, which I think it is always possible to give some positive input.

**(Mark Dunning)** Excellent, thank you! So, Emma, I have another question for you: What about Usher syndrome frustrates you the most?

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**(Emma Boswell)** Mhm. What frustrates me, I guess, is: I meet a lot of people through my work that don't know anything about their own Usher. They haven't received the right diagnosis, the right information, the right support. They might have met a specialist doctor, like Julia said. And they might have even been misdiagnosed. And it is a missed opportunity. And actually, I come up across that a lot. That is the biggest frustration.

**(Mark Dunning)** Brendan, what about the disease frustrates you the most?

**(Brendan Creemer)** I think what frustrates me the most is just the way people treat me. Either good or bad. Just people who worry about me too much. Like, there is some people who think my vision is worse, than it actually is. Like, to be honest which is really, really good right now. It is not supposed to get bad. Only much later hopefully, after the cure comes out. And, people who assume that my vision is worse than it is, try to grab me by the arm and guide me. But I don't like that. Like I just think, in my eyes it is rude for someone to grab me by the arm without notifying me beforehand.

I see myself as a sighted person and people who treat me as if I am blind is just rude. Treating me like someone I am not. Yeah, and overall, whenever people worry about me in general, like ask me, if I need help to get anywhere, or if I need them to read something for me, or something like that. That really frustrates me. I really want to be independent, that's what it is.

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**(Laura Bingenheimer)** I think, I see it more from an inner part. What frustrates me, would be that this is a progress, and that you have always this back information that you might go blind. And, so, you actually... I try to not live my life with this background information. But to live it just independently without thinking of it. That is something that is frustrating when I am thinking of it, yeah.

**(Claes Möller)** I wouldn't say like Brendan and Emma, the environment, the others frustrate me most. And in Sweden, it frustrates me a lot, that we have authorities that have no clue and don't work together. In most of the countries we come from, we definitely have resources. But we haven't planned them together to make life easier and to include all people into the society. And since speech and vision, hearing and vision is so important for our social life we should make it much, much easier to participate.

**(Isabelle Audo)** So, I am going to repeat a lot. So, one thing that also really frustrates me is, how ignorant some of my ophthalmology colleagues are. Sometimes you receive patients and patients are depressed because they were told that they - or parents from children - that they were told that they should register to a special school and learn Braille, and so on. So, which is not needed in the first place in Usher syndrome. So, it is always frustrating me to hear how ignorant and adamant some are, like: you should do this and that. Like some of my ophthalmology colleagues are towards Usher syndrome. I am also like Brendan, and what you mentioned, is that: any young

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children or adults need to be independent. And I do believe that patients with Usher syndrome can be autonomous, and independent, and very happy. And sometimes the surrounding is anxious about that and anxiety just precludes people to move forward.

**(Mark Dunning)** Good now, Kimberley mentioned before, that we need to be both: balance the positive and negative. So, when I ask you a question that you probably find strange, but: what about Usher syndrome makes you happiest? I will start with you Emma.

**(Emma Boswell)** Mhm, what makes me happy about my Usher? I think, the fact that I have managed to achieve so much, and that I have just got on with things. I have backpacked on my own. I have done-done operation Raleigh. I did Camp America. I have managed to get a degree and I have achieved so much. And that does make me happy, despite my Usher, I guess.

**(Mark Dunning)** Brendan, so, what makes you happiest about having Usher syndrome?

**(Brendan Creemer)** Makes me happy? <sup>(laughter)</sup>

**(Mark Dunning)** No one said, this was gonna be easy.

**(Brendan Creemer)** Uuh - this is a difficult question. Shh! Shh. Is it possible, you could come back to me?

**(Mark Dunning)** Yeah, of course.

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**(Brendan Creemer)** Thank you.

**(Laura Bingenheimer)** I find it very difficult, too. Of course I am trying to have a good view on it. But, yeah. It is not so easy to find something that would be making me happy. But I think, for example, at night my friends already stand there at the door and hold their arm, so that I can just grab it. And it is just a next to it thing. So, I am really happy to see that these people just see it as something that is just a part of me. But it is not a topic to talk about. And that makes me happy, that there are people out there who can do it this way, friends, yeah.

**(Claes Möller)** Without you guys with Usher syndrome I would still be a local ear, nose and throat doctor <sup>(laughter)</sup> - in Sweden. You have made me so happy, because I can travel around the world. I have met over probably 1,000 families with Usher syndrome, I work with researchers, clinicians, I can travel here without paying too much, it's fantastic! <sup>(laughter)</sup> <sup>(applause)</sup>

**(Isabelle Audo)** I would say like Laura, about what makes me happy about Usher syndrome is to realize, how some patients achieve so much. One of my patients is, - I think - he is Usher Type I, but he doesn't have Usher 1B, but another type of Usher. And he has - I think, he has four gold medals in judo at the Paralympics and I am so proud of him.

**(Mark Dunning)** That is great, Brendan, have you come up with anything that makes you happy?

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**(Brendan Creemer)** Yeah, yeah. I have got an answer. I know this might not be what you expect to hear from me. But, I really don't think there is anything about Usher syndrome that has made me happy so far. It has had only a negative impact on my life. I view it as a personal arch nemesis, maybe. I guess maybe that is a good thing to take out all of my anger on. So I end up not harming others. But I really don't think, there is anything about it that makes me happy. I am sorry to say that.

**(Mark Dunning)** That is not a surprise. That was intended to not be an easy question. I know, I have tried to ask you guys all the same questions. (man from the audience) Excuse me? Mark?

**(man from the audience)** Mark? Right here. Sorry, I just wanted to comment on that also, if I may?

**(Mark Dunning)** Sure.

**(man from the audience)** I don't have Usher, but I am a father of someone who has Usher and this question that you asked: I would say, it is not Usher in particular, that would make you happy. But it's a challenge, like many other challenges that you can have in life. And it does - I am fighting a little bit with my voice, as you can hear. But it does show that there are people around you, that there is good in this world. We have met so many friendly people after we got the diagnosis. And that - I think, many people especially nowadays, when you see, how bad things are going in the world. I that is what shows that

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there is a lot of good in the world, too. That people really fight for you, stand up for you and want to be good to you and help you. So, I think, that is beside - I mean - independent from Usher or other medical or other challenges, that is what was a benefit for us that Usher came into our family. (applause)

**(Mark Dunning)** You can't see it, Bernd, but people are taking out the tissues here. Thank you, Bernd, that was a great, great answer. So, I'm gonna ask slightly different questions of you guys. But they relate to the same sort of thing. So, for Claes and Isabelle: What is the most important thing that patients or families with Usher syndrome can do to support your research, outside of raising money?

**(Claes Möller)** I would say, that the most important thing you can do, is to organize yourself in different organizations, such as Usher Syndrome Coalition or other organizations. Because deafblindness and Usher syndrome in the scientific world are very small and you are quite few, when you are alone. Which means that the funding for research can't compete with cancer, diabetes or whatever it is. So, if you go together, it will help the research tremendously. Without you, there will not be any research. And this meeting is a perfect example of this, where we actually have gathered probably all the main researchers in Usher syndrome in the world. And we were 152 people. That is the content of researches and then we have some clinicians, of course. But that is the main thing you can do for us.

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**(Isabelle Audo)** I totally agree. And also, I think, you know, being involved in Usher, you can also bring a lot to nearly diagnosed Usher patients and families. Because, when we see them in a busy clinic, we try to take time and this is - I always imagine that it is like a cold shower and unfortunately busy clinics are like that. So, we can't review patients like the next day or the day after to answer questions and so on. And I think the families who also went through this type of things, you can really help, when we can't continue to support the patients.

**(Mark Dunning)** Thank you! So, now for my non-researchers I have two questions: We are reaching the point from a research perspective, where we are going to need to start doing clinical trials. So, my question for each of you, first, is: Would you be willing to participate in a clinical trial? I know this is a short answer.

**(Laura Bingenheimer)** Would I give money to-?

**(Claes Möller)** No, would you like to participate or would you participate in a clinical trial?

**(Laura Bingenheimer)** Mhm, that seems so far away, so I didn't really think about it so far. But I think that it depends. Because, I need to have trust in the project, and in the trial. And, I would also want to know the risks that I'm taking. But if it's promising, then I wouldn't see a reason why I shouldn't do it.

**(Mark Dunning)** Brendan, would you be willing to partici-

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pate in a clinical trial?

**(Brendan Creemer)** Good question: I might wanna wait until I am older and until the research has progressed further. But I think, if the trial sounds like it will work and I feel like I could only benefit from it, if there aren't any nasty side effects, I would be happy to participate in a clinical trial.

**(Mark Dunning)** And Emma?

**(Emma Boswell)** That would be a very, very big decision to make. And I guess, the same as everybody else has said: You would have to weigh out the risks. Because at the moment, my vision isn't too bad, and: Would I be willing to risk affecting it or changing it in some way? So, I guess, I can't give you a definitive answer. I would have to wait all up and then give my answer.

**(Mark Dunning)** I want to make a comment and I am gonna ask you a follow-up question. So Laura had mentioned earlier about that she would have to trust that she knew what is happening and trust the people that were involved. And that is a big reason, why we do these types of events. And that is, why we asked Claes and Isabelle to be here with the patients. Because we want you to know the researchers, we want you to know that you can trust the researchers. The only way, you gonna do that is, if you meet them. And that you see them as human beings and know that they care about you as an individual. And that is not always gonna be the case with every researcher.

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So, it is important that you have the opportunity to meet them and know them personally and hopefully know them for a lot of years. I have known these guys for probably 10 years now. And I completely trust them, because I have known them for 10 years. It takes time to develop that sort of relationship where you have that trust. Which is why we try and do these types of events. Because that leads me to my follow-up question for you guys: The first phase of clinical trials is a safety trial. And in most cases, the intention of a safety trial is not to improve your condition. It is to test and see if it is dangerous. So this is my follow-up question to you: Would you be willing to participate in a clinical trial, if you knew, there wouldn't likely be no benefit for you, but that it could benefit the community in the future?

**(Laura Bingenheimer)** Are you asking me? Do you already have an answer? I don't, I doubt it.

**(Mark Dunning)** Would you like to hear my answer? As I might make it a little bit more comfortable to you guys. I probably wouldn't. I would not be willing to be involved in the work and the pain that could potentially go along with the clinical trial, if it wasn't going to help me. But there are going to have to be people out here that do that, if we want to find treatments. And so, if the answer is ,no', that is perfectly fine because that is going to be most people's answer.

**(Laura Bingenheimer)** I think my answer would be ,no' because you know, you already have this disease and

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you have already had so many difficulties that you had to manage. And if you knew there would only be more difficulties probably, and I wouldn't have any benefit from it, then I would probably say ,no', because of that. Because I am already having to struggle with it and I don't want have more struggles just, so, yeah. (Mark Dunning) Brendan, what do you think?

**(Brendan Creemer)** Uh, well, for me: I am more concerned about efficacy than safety. Like, safety is - obviously is relevant. But in the long run for me it is partially irrelevant. If I want to do a clinical trial, I want to at least wait until the safety part has been cleared before moving into efficacy. So, I probably would not want to do one of those safety trials.

**(Mark Dunning)** Emma, what do you think?

**(Emma Boswell)** Well, I guess, I feel the same as everybody else. I don't know, if I would take the risk of actually damaging the vision I do have. And it would be a family decision, as well because, you know, I have young children and on the back of that, I think, I would be reluctant.

**(Mark Dunning)** Okay, thank you! Uh, yes, I asked that question because I want you to realize that it is great, we are reaching this point, where we are going to make it to clinical trials. It is going to be hard for all of us. There will be some difficult decisions for people to make to get there. And some people will have to, you know, sacrifice and be willing to do something that I don't think I would

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be willing to do. And I think, it is important for us as a community to realize that people that do that are real heroes to us. Because they are willing to do something to help the community, that won't help them directly. So, we have a couple more minutes. I was wondering, if you guys have any questions, that they would like to ask anybody on the panel, or the panel in general? Any hands? No? Oh, we have a hand over here.

**(women from the audience)** Hello! Is there an international patient register existing where you can register yourself and your mutation you have, because there is one in Germany, but, maybe it is also interesting for the whole world?

**(Mark Dunning)** That is an easy answer, Nancy, would you like to stand up? So that is Nancy O'Donnell over there, and she runs the Usher syndrome coalition international registry for people with Usher syndrome. If you go and find Nancy after the symposium, she will be happy to get you registered and it is actually, there is actually, a German page on the registry. You want to add anything, Nancy?

**(Nancy O'Donnell)** That's perfect, come on over. I have my laptop.

**(Mark Dunning)** Anybody else have any questions that they would like to ask?

**(women from the audience)** Hi, I am Anne Schuer, I am parent of 4 kids, two with type 1b, as a mom I am won-

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dering if the people on the panel who are patients could speak about things that their parents did, that they found supportive and maybe things that were not helpful, so that I can replicate or not replicate. And if the researchers could maybe talk about effective parenting that they have seen with their patients and families. You know, even talking to our young children about this diagnosis and teaching them a healthy way to cope with whatever feelings they might have as things change. I think that for me is my greatest need, watching my children go through this, is, how do I be a support and not hover too much. And you know, I don't know, make sure, that they can become the people that they are intended to be and to not let this diagnosis be something that completely defines them.

**(Mark Dunning)** Good question, Emma would you like to answer that first?

**(Emma Boswell)** My mom and dad are divorced, but I have a set of step parents. My mom knew I had Usher at 7 but didn't actually tell me until I was 18. It was when I was going to operation Raleigh and I started scuba diving and I realized that my balance was very bad, and I asked my mom about this. And said, what is wrong with my vision? What is wrong with my balance? That is when she sat me down. My older sister also has Usher, my brother doesn't. She sat all of us down and told us, that me and my sister had Usher.

But after that point, I know it was rather late, my mom

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actually encouraged me and my sister to be very independent. My dad on the other hand was very, very different. He didn't want me to go travelling. To Camp America, he didn't want me to do the things I wanted to do. I actually think that was because I don't communicate that well with him. I lived with my mom, grew up with her and I communicate with her amazingly.

Actually the one thing I do wish, is that both of my parents could sign, because I don't know what my vision is going to be like in the future. I might need to use hands-on signing, that is why I've taught both of my children to sign, because I want to be able to communicate with them for the rest of my life. That is probably the biggest thing, I wish my mom and dad had learned sign language. I am very open with my children as well, so I would say that is definitely a bonus, a plus.

**(Mark Dunning)** Brendan, do you want to answer the question?

**(Brendan Creemer)** Sorry, what was the question?

**(Mark Dunning)** The question was, what things have your parents done, that have worked well and what have they done that has not done well and your dad will cover his ears!

**(Brendan Creemer)** Sorry, what was that?

**(Mark Dunning)** Do you have an advice for parents as to

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what to do to make it easier with kids with Usher syndrome.

**(Brendan Creemer)** Definitely being positive from the start like saying: Having Usher syndrome won't stop you from all the things you like doing right now. That obviously helped me. As soon as they are old enough to understand what is going on will all this research, maybe you can tell them about it, and be positive about it. Say like: we are working towards a cure and we hope to have it out in the next few decades. Just being positive, telling them regardless how bad this seems, they can still live out their normal lives and be happy.

**(Laura Bingenheimer)** I agree with you. And I think it is very important to be honest. For example my mom, she told me directly what I have. She was not very enthusiastic about it, of course, but she just told me like, „You have this disease called Usher syndrome“. I was like, ok, and then that was it. We were very honest about it, that was very important. But in the end I think it is important that the parents listen to the child, when it says something, but don't really do something more when it doesn't say something, because I think that is - I would find it restricting if my parents would contact me and be like, „Yeah, what is it like for you?“ and things like that. This would make me feel being different and, I would have a problem with that. So, what my parents did right was not doing too much.

**(Claes Möller)** My experience is that the - I have seen now

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over a hundred children who now are grown up, they constantly say to me, that those who come out the best, are those, where, - as you said - where the parents told them very early about the problems that they might face. Also especially when you are young, very young kid, tell them about if you have Usher type I, that yes, you are a little bit more clumsy, you can't do everything. But that is just because you are deaf in your balance organs. So that they don't hear that from others. Then educate all the other caregivers in the pre-school etc. Because many children will get very stupid comments from ignorant people.

**(Mark Dunning)** Anything to add, Isabelle?

**(Isabelle Audo)** Yes, I can only speak also from my experience. First, I think as parents you need to trust your children - any children, not only your children with Usher 1b - that they will be going to be fine. And second, I must admit, - except of course when you do any RG on the 6 months year old - but when I see patients, children with Usher syndrome, I tend to see them with their parents. And I try to explain in a simple way why they came, because, you know, performing RG is not something easy. And, and try to explain them, you know, you don't see very well, maybe at night, and so to explain them why they don't see very well.

And that very simply, because, maybe I am wrong, but I do think that maybe not saying things can create false anxiety and just saying: We understand why you don't see well at night, you are going to have a flashlight and everything

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will be ok. So, I do think being honest and rationalizing things help a lot, rather than hiding things. Because the kids, they know. They understand quite a lot, I think.

**(Laura Bingenheimer)** Can I add something? I think the diagnosis effects - this is the first time you actually have to do something with the disease. I think it is very important to think about how you tell your child. I think it is very important to just say it without any emotions, so that the child can like put it in the right order. Because I think if you would already tell the child with a crying face, then it is probably, not the best thing for the child. Because it thinks, „Oh, my life is over and this is probably going to restrict me so much“ and thinking bad about it. I think that would be bad. Yeah.

**(Mark Dunning)** Great, thank you guys, so, we have gone over our time. But I know, Brendan, wanted to say a few words, do you have...

**(Brendan Creemer)** I had a speech that I wanted to give.

**(Mark Dunning)** Sure, do you have to time to give it? Do you have it with you? Are you ready to go?

**(Brendan Creemer)** Yeah.

**(Mark Dunning)** Hit it, kid.

**(Brendan Creemer)** Alright, you heard a fair amount about me earlier when I was answering those questions, but I

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think it is time to tell you my story, to really inspire people to take action. So, once again, you know my name is Brendan Creemer, I am 19 years old and currently in college. I go to the Lewis and Clark in Portland, I am from Palo Alto, California. And over these past few years I have taken some action to raising awareness for Usher syndrome in my community, so I am gonna speak about that. So, with that, here I go. Dear fellow community members, Usher families and researchers, imagine for a moment that you are a child who has recently been diagnosed - no, that your child was recently diagnosed with Usher syndrome.

You are probably between 9 and 15 years of age, living a pretty normal life so far. You go to school, hang out with friends, play with toys and have fun. You are simply cruising through life, nothing will go wrong, right? Then one day, completely out of the blue, your parents tell you, that you have Usher syndrome and that your eyes do not work as well as you believe they do. In that moment you feel like your whole life was a lie, you are devastated, you isolate yourself from your friends. You give up all of your big life goals feeling like there is no way out of the dark future that looms ahead.

Well, I will tell you this, I am NOT that kind of person. Instead I am a person who upon realization does not see Usher syndrome as a thread. I am a kind of person who takes action! A person, who reaches out, who is motivated, who is resilient, who knows that there is some solution out there, regardless of how awful this situation

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seems. Let me tell you a story, a story of how I found out that I had Usher syndrome, stayed motivated and eventually won several victories in the war against this dreadful disease. I was diagnosed when I was very little, but my parents withheld the truth from me until I was 10. For the next 4,5 years I dismissed it, not denied, dismissed. Thinking it was not going to be an issue at all, because I was pretty happy with the way things were going for me at the time.

In the middle of 9th grade, however, I became more aware. I was upset with the intense accommodations I was receiving in school at the time, because I had grown out of many of them. At the same time, I learned about biotechnology in my biology class and had just learned about genetic mutations. It was in that moment, when I realized that all of my current problems could be traced back to a single mutation in my DNA. Immediately after that I decided to pursue biotechnology as a career, while swearing a personal oath to cure Usher syndrome before I lost too much vision. In the meantime, I would raise money for the research that was currently going on in order to give as much support as I could.

During my sophomore year, I began my own personal war against Usher syndrome working together with this organization called the 'BrankstreetGarage Fund', now re-named 'Think Fund', an organization in Palo Alto, California, where I am from that helps teens run programs. I got together a group of people and planned an open mic talent show event where all the proceeds went to Usher

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syndrome research. I raised over 700 dollars for Edwin Stone's research at the university of Iowa. Additionally, I was able to spread awareness to the community. We named our group ,science for sight' and continued to build more fundraisers the following year when I was 16. Unfortunately, I had to shut down the club at the end of the year, because we weren't having as much as success as before and the rest of my group lost interest in the project, but I did NOT give up hope.

During that time I was getting more and more involved in another organization in my Jewish youth group, BBYO, I don't know if any of you heard of it, it is pretty popular, a great organization. Anyways I got pretty involved in it at the time and saw another opportunity. At the start of each semester, the chapter, the division of my youth group that I was part of would choose a new standup cause, that is a Jewish organization we would spend the entire term raising money for.

Since Usher syndrome Type I, the kind I have, is very common in Ashkenazi Jews, I decided that would be a perfect idea for a standup cause. Together with my chapters of the vice president of Judaism and community service, we made Usher syndrome our standup cause. The guys in my chapter, which, I would like to acknowledge, my chapter was called ,Simon Wiesenthal, AZA 2524' of the region center. Region west number 45 of the BBYO, great organization that.

Parents, if you have a kid who have Usher syndrome and

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if your guys are Jewish, please try to get that kid into BBYO, that would be a great way for them to raise money and awareness. The guys in my chapter worked tirelessly throughout the winter 2017 term, the second half of my senior year, to raise money for the cause. We created a GoFundMe page and after hosting a marathon towards the end of the term raised almost about 2,000 Dollars for Usher syndrome. My senior prom graduation occurred soon afterwards, so it felt like a real victory to me.

Right around that time, I heard that the mouse model for type 1F have finally been produced, which only further strengthened that victorious feeling. Nowadays I am in college, simply working my way towards a degree in biotechnology where I can tackle this disease head on, but I can't do it alone. I will need your help, with that I have one final message for all of you.

To the people close in age to me with Usher syndrome, high school kids, college students, young adults, teenagers: YOU have the power to change things! YOU have the ability to reach out to your community to spread awareness and to raise money! YOU have that choice to either give up and assume everything is helpless or to choose to take action and not only help yourself but others around you as well.

You have that potential inside you to become a game changer. One who motivates and inspires those around them in ways never seen before. YOU have that potential to do amazing things. To the families, especially those of

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very young children. You may not see this, but your child has the potential to do great things. Your children have the potential to become famous, almost as if they were the next Albert Einstein or John F. Kennedy, if they are older. NOW is the time to start encouraging them. If they too young, simply encourage them when they are older, also if they are Jewish, encourage them to join BBYO, if they are not Jewish encourage them to join community service groups at school or in their community to take action. Spread the message of how I took action instead of giving up in order to help others as well as myself. Inspire them to make their own impact on the world. Motivate them to be game changers, to join me in my quest to cure Usher syndrome.

To the researchers, thank you for your, all of your hard work you have done so far, and I wish you the best of luck continuing the study. To the families, thank you for supporting your children, as much as you have, and I hope you continue to support them further. To those with Usher: DO NOT GIVE UP! YOU have that power to change things, use that to your advantage. Community members, families and researchers: Lets eradicate this disease. Thank you. <sup>(applause)</sup>

**(Mark Dunning)** Thank you Brendan, a good note to end on. So, this is the end of our conference today and the end of three days for all of you researchers. I really want to thank all of the researchers who stayed around for today, not only to give speeches, but there have been a number of researchers who have attended this confer-

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ence and have been available for people to talk to. We really appreciate that. I know we didn't have a ton of time to ask questions at the panel, but we are all going to be together at the family dinner hopefully, and that is a great opportunity to ask questions there. Irmgard, is there anything else I need to say? There is? Or no? Do I need to say anything else? You are getting a microphone.

**(Irmgard Reichstein)** Yes, I would like to thank the technical team, I think they did a very, very great job. And even the interpreters! (applause) To be honest I never saw so good interpreters like we have had today. Because they have been images, they have been so fast, especially now at the end. Thank you very much to the interpreters!

(applause)

**(Margaret Kenna)** Just on behalf of the organizing team, we knew how hard this is, and there is one person. I don't know if she is here, with a 6 months year old baby, Krista Vasi, who works for the coalition, and who remembers not only everything we remember, she remembers everything that we don't remember. Thank you! (applause)

**(Mark Dunning)** Uwe, would you like to wrap this up?

**(Uwe Wolfrum)** I guess I would like to thank you for coming to Mainz. It was a pleasure to us, the organizing committee, Sebastian, Kerstin, my wife, and all the co-organizers from the US, from Austria, all over the world, more or less, what we did here. I guess you, I hope you enjoyed maybe also the boat cruise yesterday. We all - we organized also the weather. This was also something, when

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we can do such kind of research, I guess we really, really can forward a treatment for all of you. Yeah. Thanks for coming. Yeah. And I hope, that we will do our best in research. We are researchers at the university of Mainz and we hope that we can help you. And I like to thank Krista, is she around? She was organizing with us with, I guess, most of the time on the phone and the phone conferences. I don't know, maybe she counted this, maybe around 50 conferences we had to organize this here. Maybe I can hand to you, Sebastian, who was in charge, more or less, for the patient day to maybe give some words.

**(Sebastian Klaes)** I don't think that I need to say quite much more. I have enjoyed this symposium and, yes, I hope we will have a good talk tonight and maybe, yes, I hope to see you or most of you in the next symposium, I don't know where.

**(Uwe Wolfrum)** I would like to thank the sponsors as well, you always saw these slides and of course it wouldn't have been possible without the support from the sponsors to get, this meeting together. Thanks a lot for coming and we will see you. <sup>(applause)</sup>

For the patient dinner we will meet downtown Mainz and it is quite easy to get to the place. You will find the address in your booklet and you can go by public transportation or by taxi, you have to indicate where to go. This restaurant will be very close to the theatre. And there you may get also around. The restaurant is called 'Haus des Deutschen Weines', it is the 'house of German wine'. And