Usher Syndrome Data Collection Program Webinar with RARE-X

November 16, 2022
Transcript:

Hi everyone! Welcome! I'm Krista Vasi and I want to thank you all for joining us today. We are so pleased to share with you our exciting partnership with RARE-X and the launch of our new Usher Syndrome Data Collection Program.

The Usher Syndrome Data Collection Program is how we as a community are taking an active role in research by providing the robust data needed to fully understand Usher syndrome. The Usher syndrome DCP will become part of the fabric of our community, essentially a living natural history study.

The USH Trust is how we connect the community and together, synergistically, we will speed the search for treatments for Usher syndrome. This is a perfect illustration of the Coalition's role in bridging the gap between the community and researchers.

The launch of this new program is the culmination of years of advancements in research in how data is collected and secured and conversations with you, the experts, living with Usher syndrome.

With that said, I'm so happy to introduce our speaker today, Megan O'Boyle, who is the patient engagement lead at RARE-X and she will explain in more detail what this data collection program means for you. Thank you, Megan.

Thank you, Krista, and all of you who have taken the time to come today. I'm so excited for your community. This is a huge, huge step forward for any rare disease to have this type of effort in data collection and I'm really excited for you, and you should be proud of your community for being such forward thinkers. I'm going to go ahead and share my screen and present slides, and we will have questions and answers at the end. I believe there is a place to put questions at the bottom of your screen and hopefully, everybody can...um uh, access...see that I have slides up.

So today we're- I'm going to go over who RARE- X is, um, and what we're trying to solve for and how we're going to keep your data safe, and how you're going to get started to enroll, and some other tips that we've learned from other disease groups along the way.

So RARE-X is a non-profit and we were created to accelerate research and treatments by removing barriers for organizations like yours who were left with the responsibility of learning everything about data collection, and researchers, who were having a hard time getting access to high-quality data. We are a platform that collects data, which we'll talk about today. We have the ability to connect to existing data that might exist somewhere else.

And I know there are other data collections in, um, the vision loss research world as well as the hearing loss research world that may be potential data sets that we could connect to later, and, most importantly, we share, or disseminate that data with researchers around the world.

When I talk about sharing data today, I always mean de-identified data. The only entity that will have access to the identification of participants is RARE-X. Your organization will only see de-identified summary data and the researchers will only see de-identified data on the participants. We do not own the data. None of the patient organizations in the Usher community own the data. You, as the participant, own the data. We do not sell the data, not even to pharmaceutical companies and Biotech Industry.

So, as Krista mentioned, this has been a long shopping process. Your community has left no, um, stone unturned in terms of looking at the various options for data collection and they, after several years, landed with RARE-X. And we asked them why they chose us, and one is that you will own that data - you as the participant, not them; that we have the highest, um, standards for security and privacy; that there are no costs to your organizations and there are no costs to researchers. So all researchers - from academic researchers to industry - who are pharmaceutical or biotech researchers - all have equal access at no cost to this data. Again all data would be de-identified.

The fact that RARE-X collects mostly structured and standardized data was very important to your Usher leadership. Standardized means that we are searching for the best standardized, validated, vetted, existing surveys for the symptoms that we are asking about. So we did not make up the surveys that you will be answering. We have had, uh, outside advisors landscape all the available surveys and choose the one that will yield the best data for research.

We ask about all systems of the body for two reasons: one - you may have several or more than one disease, and so it's important that you report on everything. Do not second-guess yourself. If you have a symptom and you don't know if it's related to Usher it doesn't matter. You should report on everything that that you feel medically

and the other reason that we ask about all body systems is some diseases are not fully characterized that researchers have only asked about certain systems and they've left out others and so it's important for us to make sure that we know all of the symptoms that the Usher community is experiencing.
One of the greatest things about RARE-X is how we have streamlined the access of researchers to this de-identified data. There will be more eyes on this research from around the world. All of this is to speed up the development of treatments and as I said we will eventually be able to connect to other data sources if they are of value.

What is the benefit to you - you are busy you have answered other surveys and you haven't gotten much for your effort in the past so 1) you will see data returned to you. We will give access to your leadership to de-identified charts that show summarized data so nobody will know that it is your data but you will, there will be percentages - how many people from certain countries have enrolled, how many people are experiencing specific symptoms.

It is a great way for participants that are interested in being included in other research studies outside of RARE-X or clinical trials to be found so researchers can go into RARE-X and query based on genetics, symptoms, or disease. So if a researcher were to go into RARE-X now we have 40 diseases and several have a vision impairment of some sort that researcher if they were to query on the vision impairment question they would get participants from various different disease communities and this is very helpful if they're looking to develop treatments based on symptom and not disease.

As I said there will be more eyes on your data. Researchers from around the world will know about this data collection even if they don't know about Usher syndrome because it will be highly publicized by us.

You can go in - and we encourage you to go in - and update your symptoms at any time. This is not a study where you only can participate once a year, so we want you to think of this as not a research study or a favor to the research world. This is for you, by you, so that your community can better understand what this disease is, how it develops, and how it affects families.

You can manage your data and how it can be shared so it's up to you whether you want to share your data broadly or if you want to put limitations on that.

And finally, all of this effort will and should speed up the development of treatments and devices so at the top of this busy slide you'll see several different diseases and as I

said we've welcomed over 40 or 45 diseases so far and we'll continue to bring them in. There's over ten thousand rare diseases and they're all welcome in RARE-X. Each disease has a front door or Community page and we will be giving you the link to that today.

There there are frequently asked questions and other information that you may want about this data collection program.

Once you hit get started you will be in what is on this slide - the middle rectangular green box - this is the data collection and everybody that comes through RARE-X will follow the first initial steps of consenting, deciding how they want to share their data, providing their demographics and general information, and then their first survey will be the health and development survey.

This is what we call the head-to-toe survey. It asks you about all systems of your body.

From there, additional surveys will, may be added to your dashboard which is represented on the right of this slide with these smaller uh surveys.

At the bottom of the slide, we have the important researchers - again they could be from academic centers, medical centers, pharmaceutical companies, or biotech companies - and these researchers will go in one door, provide their credentials, agree to the data sharing agreements, and then they can query on the data.

On the left here, you will see that the traditional way of sharing data with researchers is one at a time. A researcher knows about your disease, they hear that there's some data, and they ask to see it, and then that organization has to de-identify the data and share it with the researcher and then wait. and maybe...[lost audio]

How RARE-X is doing this - many diseases are putting data into RARE-X and many more researchers will be able to access that data and compare data across diseases. With that example I gave earlier, where there are various diseases that may have the same symptoms.

So what do you need? You need email and a password and you should only use the approved browsers which are Google Chrome, Apple Safari, and Microsoft Edge. You don't need to finish this all at once.

I would encourage you to enroll, upload your genetic report and get through at least the health and development survey at your first sitting um but these will save automatically and you can come back and finish any time.

We will send reminders if you have started a survey and you have not yet completed it and remind you to come back. So you'll get the link to the Community page today and it will be shared widely over social media. You will set up your account and like many things nowadays there will be double verification where a code will be sent to you.

You will answer your informed consent. If you are a parent of a minor, you will consent for yourself and decide how your data will be shared and then you will consent on behalf of your young child and then answer how their data will be shared and then you will you will answer surveys on their behalf.

This is the beautiful Usher Community Page. As you see, it says "Usher syndrome" in the upper left corner. This is for all Usher patients/participants around the world. It doesn't matter what organizations that you heard about it from, it doesn't matter you know and currently it's in English we will be adding other languages in 2023.

This page has frequently asked questions at the bottom and information about what the data collection is. As I said when you hit "Get Started," you will be in the RARE-X in the (excuse me) data collection platform. under the get started button, there is um a link to our terms and conditions um and you excuse me our privacy policy and terms of use, if you're interested.

The first page that you'll get after you join or get started is to determine what consent that you are sent whether you are a caregiver of a minor child and so forth so if you could just answer these basic questions. I just want to note that all biological parents of underage children with Usher syndrome would also be considered carriers and should also choose patient participants so um on the right, you'll see that there's different roles: patient participant, caregiver participant, and a person who has lost a loved one. You can do any or all of those roles, depending on your experience.

I want to take this opportunity right now to apologize to your community for the use of the word patient. We understand that most people do not consider themselves patients. Unfortunately, because we are serving all rare diseases and many have caregivers of adults and they are both the participant and the caregiver we had to differentiate somehow and so we were forced to use that word, so we apologize.

As I said, there'll be account creation um where you know set up your password verification code I'm going to go quickly through these as I think you're um familiar. Here's your login screen so once you leave the data collection program and come back to update or finish, this is the screen that you will always go to. You can go to the Community page and hit login which is next to the get started button. Again, you can access our privacy policy at the bottom of this login screen.

The next screen you'll see is the terms of use for Matrix which is the technology platform that rarex is is powered by and at the bottom of that once you hit accept you will be then taken to the informed consent form. We tried to keep this clear and transparent and simple to understand so it's in a question and answer format and we have tried to keep the legalese out of it.

If you have any questions you can always contact the principal investigator who's in, listed at the top. There are several pages to the consent. When you get to the second to last page, you then again have to to specify what role you are, if you're a parent or caregiver, legally authorized representative, a um, the person who has the disease or so on. You will enter your personal information. Again, this is only information that RARE-X will see.

This is your contact information, um and these demographics in general information. All of our surveys are optional but all of the questions within the surveys are mandatory because we, researchers don't know how to value a skipped answer but there are answers such as prefer not to answer that you can use.

At the bottom of the general information screen you'll see, are you a person with a rare disease and you want to just start to type in Usher and it will show up on the drop down menu of diseases if you happen to have another disease go ahead and click on other and you can tape, excuse me, type in what that disease is it doesn't have to be related to Usher.

We want to know everything about your health. Perhaps you may qualify for a study based on a different condition than Usher.

This is where you decide how you want to share your data. There's only a few questions and the first page, you have to choose one you want to share your research, excuse me, your data for general research, which is any kind of research including research about age race and ethnicity or studying traits of certain populations or you can choose the second option which is more limiting but perfectly acceptable and that's

just research being done in areas of health, medical and biomedical research. And these options are explained in detail on those pages the second page is completely optional. When I enrolled I chose not to click either box.

The first box says that you will share your research with all researchers except industry, so that means you do not want to share your research, your data, with researchers from pharmaceutical companies or biotechs. The second question asks about sharing your research with only researchers that have gotten institutional review board approval. That means if there are academic research their institution has reviewed their study and given them a stamp of approval. Again, these are limiting but they're perfectly acceptable choices. You can change your answers to any of these at any time. So you can go back into the data sharing preference survey and put more limitations or remove limitations.

The rest of the questions for this survey are a poll to help us communicate to your community: who is already in research, who is willing to do other types of research, so we ask whether you have bio samples somewhere, and if so, where are they. We aren't currently connecting to medical records but if that was a possibility, would you be interested? So you're not signing up. Unfortunately, I can't make that happen that easily but it would be important for your organizations to know if there is an interest so if they decide that that is an investment they want to make.

Under other possible data sources, we're asking if you've been in other research studies and if so if you can just tell us what you remember about them, either the location or the researcher.

Toward the end of this survey, you can choose to share your name and email, it says contact information. It's just name and email with any of the patient advocacy groups that are partnering with RARE-X and there are several um so this is just to share your name and email so that you can be included on their communications. They will not see your medical information.

The second question is that you want to you want to be included in summaries on the dashboard. This means that when your organization um can access summarized identified data that you would want to be included and I can't think of a reason why you would not. If you have more than one person with Usher that is being enrolled and, for instance, if you are a caregiver and you have a child that is a minor you would enter your information and answer your, those two surveys I just talked to you about, and then hit add patient.

Once you have two people, when you log back in, you have to choose which person that you're working on, and if you don't see surveys on the participant that has Usher, you you should click on this, it looks like a book with a plus sign on the far right corner of your dashboard. And the add patient button again is to add an additional person. If you leave and you forget who you are entering information, the left upper, excuse me, the upper left corner will indicate the name of the participant that your um, whose dashboard that you're in.

So once you get through the consent and the data sharing, now is the fun and exciting part. This is where you answer questions about your health. The first survey we want you to go to is the Health and Development Survey and the last survey we want you to go to is the Diagnosis Survey because the Diagnosis Survey is just to report what you have not yet been asked about. So it's free text and I'm concerned if you go to that too soon, you're going to enter information and then find out later that you're being asked in those other surveys.

So complete the Health and Development Survey, it says that it takes 15 to 20 minutes. We've never seen that to be the case but there are 43 questions. You do not need your medical records. It asks general questions like does the participant have breathing or respiratory issues? Yes, no or unsure. Does the participant have ear or hearing issues? Yes, No or unsure. So it's very simple.

Again, it doesn't matter if the symptom is related to Usher. If you have a symptom, or you think you may have a symptom, go ahead and answer. So answering yes or unsure in this survey will add an additional survey, called a Level 2 survey in your dashboard, if we have one. I want to point out, up in the right hand corner of this, of each page, there is a red "X". You can click there to save and exit. If you exit without clicking it will save the last completed page so you will not lose data. Toward the end of the Health and Development Survey, there are questions about whether you have genetic tests, and if so, we'll ask that you upload them and I will show you how to do that in a moment.

Once you've completed that survey, I direct you to the documents item in the left menu and when you click on that you just hit upload, and upload your scanned genetic report. So for every yes or unsure, if we have another survey, it will be added to your dashboard and it's shown here under Level 2 surveys and studies. Again, each one says that the amount of time and how many questions.

At the top of your dashboard, I want to direct you to a gray box that says completed. So if you can't remember what you have and have not done, your completed surveys are in one tab, and your new or In-Progress surveys are in a different tab. You will go back to your completed survey if something changes. For instance, if you don't have cardiac issues and you develop cardiac issues a year from now you would go back to the question in the Health and Development Survey and change your no to cardiac symptoms to a yes, when you change that, if we have an additional survey, it will be automatically added to your dashboard.

New surveys will come out on a regular basis so you will find out about new surveys if they're related to your previous questions through email. Or when you log in at least once a year, you may see that there's new surveys that have been added. This is the example of the Eye and Vision Level 2 survey. it's still high level, it's not very detailed. It asks about, I think 10 questions. If you say yes, um you then have to indicate what the age of symptoms started and then the age of diagnosis. This could be the same day or year but it could also, there could be a gap. So there could be a time where you had a gut feeling about something but your doctor said no no, I don't think that's it, and then the age of diagnosis would be when somebody finally gave you that diagnosis in your medical record.

So this is the remaining questions on Eye and Vision. If there's something you want to report and it was not asked then at the bottom of every Level 2 survey, there is a question that says, you know, if you have additional issues and then if you click yes, a text box will open and you can use free text. Please do not use any personal names in the free text because that's an identifier we don't want. You can add as many additional issues as you want to each Level 2 survey. This is just an example of the Ear and Hearing survey. Again, these are brief.

There is a additional, um there is a neurological survey, there's a survey for every symptom of the body, pardon me. This is an example of the neurological survey. This is the survey I said to do last, this is the Diagnosis Survey. And this is, you know, if you have more specific genetic information you want to provide or if you have something that none of the other surveys has have asked about, you can report it in this diagnostic survey. Again, I would do this one last.

We have a Quality of Life survey and some of the questions will ask about the last 30 days, some will ask about the last seven days, and some are more general. Your questions are for research. It is not to um, if you're reporting depression or anxiety, this is not going to route you to an additional resource. And again, all the Quality of Life

survey is optional, but all the questions are required. There is a prefer not to answer or an unsure.

The reason quality of life is so important to uh pharmaceutical companies and drug developers is that it's important to know what the status of quality of life was before treatment versus what it might improve to be.

So we're back to the dashboard. If you need to make a change, you click on the three buttons or three dots to the far right of your screen and you will see View, Edit or Email. You can email your surveys to yourself or anybody you want. You can share your data with whomever you want. HIPAA has nothing to do with that. You can edit, which means change your answer, or you can just simply view to see you know how you answered something.

So how is this different from other things that we've participated in? um and I may have a co-worker coming in... [Background noise] So, this is comprehensive across all body systems, and this is your community's data. It's not the NIH data, it's not Dr. Smith's data, um it is not an unlimited grant, it is the Usher community's global data collection and it's only as good as the people who put in the data so I encourage as many of you to enter your data and update it as often as you can.

This is for cross-disease research as well as single disease research which as I explained earlier is really important for treating symptoms. We're going to be returning de-identified summary data. Most research studies are not able to do that so that is a diff- another difference.

We are here to complement these other studies, we are not replacing studies that are taking place. If a new study comes along, we would encourage you to participate in that if it's something that interests you. So we are here to help those researchers find appropriate participants. So we are not in place of any research.

Again, you will have the ability to update this at least, you know for years and years to come, and we would ask that you do it at least annually.

On this final page, I just wanted to direct your attention to a link that was shared, which is www.perkins.org/accessibility-statement/. This is a resource from the Perkins School with some tools that you may want to access if you don't already use accessibility tools.

If you have any questions, experience any technical issues, or you want to provide feedback of any kind, please email us at support@rare-x.org and please CC n.odonnell@usher-syndrome.org. We want your community leaders to know if you're happy or unhappy or um what feedback you have. So we would like to know how we can help you and we would like them to know how many people are requiring assistance.

Thank you so much for your time and your attention and I'm going to stop sharing slides and take questions.

Krista: So, this is Krista speaking. Megan, is it helpful if I read some questions to you that I'm seeing in the chat?

Megan: that would be great, thank you.

Krista: Okay, a couple people have already asked where they can find the surveys so I have shared in the chat the live link to the Usher Syndrome Data Collection Program. This is the program, this is the platform so we're looking for, you know, this is where you enter your real data. Everything is live and ready to go.

So one question we have from Enrique is, "Could we as users get in contact with other community members who want to share their data, for example with someone with the same gene mutation?" Megan, I can probably answer part of that as well.

Megan: That'd be great.

Krista: Yeah. So because of the privacy and security of the Usher Syndrome Data Collection Program through RARE-X, there won't be any way that you'll be able to identify other members who are, other individuals who've entered their data but that's where what I mentioned at the start of this meeting, that's where the USH Trust comes in.

The Coalition will still maintain and expand the USH Trust, which is our ability to connect the community. So if we are contacted by an individual who has a diagnosis of "x" you know, type of Usher syndrome or a specific mutation and is looking for others that they can connect with we, myself, Nancy O'Donnell, Julia Dunning, we work on a daily basis really to make those one-on-one connections so you'd come to us for that. Megan, do you want to add anything to that or…

Megan: No, No.

Krista: For Megan, Catherine asks "How often are dashboards updated?"

Megan: So the data is always live. Um, we add new uh surveys as our working groups um have them so we will be releasing uh 13 new surveys associated with neurodevelopmental disorders in December because that's the working group that just completed the work.

We will have a vision loss working group and a hearing loss working group and a neuromuscular working group and so forth. As those best surveys are identified they will be added to dashboards if you answered a question associated with that survey.

So it's important that if your symptoms change that you go ahead and update that original health and development survey. We make updates monthly to the to the technology and the content. So for instance, we got feedback from a group that wanted migraine to be given as an example in neurodevelopment and so we'll be making that edit and it will be released next month. So, um, those kinds of improvements happen monthly.

Krista: Thank you. And I see a note that my volume is a little low and I'm hard to hear but Megan you're here, you're heard fine. So I'll try to speak clearer and louder let me know if there's still an issue.

Another question we have from Mariah, "What is the biggest benefit of this research for the participants?"

Megan: It depends on your interest in research. If you're not interested in research or you're not interested in, you know, treatment development, you will benefit by just knowing more about Usher syndrome because as the data comes in, we will be able to share the summarized results with your organizations.

So um, you know, for those who feel alone in some of their symptoms it may just be that nobody on Facebook or nobody in social media is talking about a particular symptom and maybe in fact it is more common in the Usher community and so getting aggregate data back I think is a very big benefit.

I feel that it makes you a much better, more empowered advocate in your medical team and in, for some people, in their education teams. Not everybody believes a parent or

crazy mom when she asks for something but they will believe a chart so having data that you can take back to your, like I said, your medical team to um show what you know common symptoms are or um other information it can be helpful.

If you're interested in research, it's a very easy way to be found for clinical trials and studies.

Krista: Thanks, Megan. This is Krista again. I see a couple hands raised as well so I can call on Keith Hodder, um to I'm going to allow Keith to speak and ask his question and, Keith, you should be able to be heard now. Okay. There we go.

Keith: Hello?

Krista: Yes, we can hear you.

Keith: Okay, thank you. I had to make sure I unmuted correctly. Um I have two quick questions and um, and because I'm legally blind obviously here um, I noticed that you put in this chat the website to go in and put the real data in. Um is that part of the Usher Syndrome Coalition website? I can get in through that uh platform or is there a separate web address um that we need to um click on to get into the RX?

Krista: Thanks, Keith, this is Krista. So we do have, on our website we have um links to that web page that I shared with you. Okay. We have a spotlight on our home page, we have a data collection section of our website now that will direct you to you to this. So if you get lost, if you lose the link that I've shared today, it is on the, it's a RARE-X link.

Keith: It's a RARE-X link. Okay, so is it like rarex.com slash something or is it okay just...

Krista: It's ushersyndrome.rare-x.org.

Keith: Okay all right so Usher syndrome I'm sorry yes yes sorry rare hyphen X .org Okay great. Okay, and the second question I have and I'm sure this has been covered but I just want to make sure um, I use um voice over on my iPad um are all the features within the website and RX's web- uh RARE-X's website um accessible through the voiceover feature, in other words, will all icons be identified because I find problems in some people's websites that when you use voiceover it doesn't always identify tabs and buttons and whatnot that you need to click on to do certain things just like the uh presenter here was telling us through the dashboard.

Krista: Megan, I know you can speak to this a bit because I know you've been um working towards this do you want to…

Megan: So we are making our best efforts, and I will not say that it is perfect. We did have this um platform evaluated by Miami Lighthouse who does that as a service and we acted on as many of the changes as they recommended that we could but there is room for improvement. (Keith: Okay.) We are happy to get your feedback. (Keith: Okay great.)

For instance, when we launched to another vision um loss disease, LHON, they informed us that the terms of use was not could not be seen by JAWS, read by JAWS. So we we made that modification and now it can.

It's important if you have issues that you do report it to us at support@rare-x.org and copy Nancy O'Donnell on that so she can keep tabs.

Keith: Great, okay great. Thank you very much for the update and I think this is a great program. I'm very interested in getting involved with it. How is it different from the Foundation Fighting Blindness uh Retina Tracker?

Krista: This is Krista speaking. Um so we will always say, we will always support, you know, your participation in My Retina Tracker and any other studies or registries you've already you know participated in. (Keith: Right.) This is, this is different in a couple ways, um because that Megan has touched on, in terms of looking at all body systems but also uh being able to hold a place for Usher syndrome specifically. Right um and, you know it's going to be an ongoing, it's not going to be a one-off, you know, you answer your, put in your information one time so we do right right, the potential here for that natural living history, um a natural history study. Megan, do you want to add to that? I know you speak a lot to a lot of communities about you know everyone's been asked to put their data in so many places.

Megan: Right and we don't want you to have data exhaustion but very rarely is any one thing a you know serves all the right purposes so um we may not go as deep dive on some things at this point as a researcher doing a specific Usher um syndrome might but they may not be asking about other body systems for instance. (Keith: Okay, alright.)

I don't want you all to be too frustrated if you don't get asked questions that are more specific about your symptoms associated with Usher it just may mean that those questions would be in a level three and we haven't identified those surveys yet. So as I said, there, we'll always be adding surveys.

Keith: Okay, great. Alright, well thank you very much for the update and I'm looking forward to participating in this.

Megan: And we look forward to your feedback as well Keith: Okay, great no problem. Thank you very much.

Krista: Thank you, Keith. Yeah, you're welcome. This is Krista again. I'm going to take a couple more questions off the Q&A and so we have um a question from Megan: "If you are not a minor and your parents are still living should you get them to put in their information?"

Megan: That's up to you so if you're not a minor you would enroll yourself and um if they are deceased and you think that there's an association with your health, you would enroll them as a loved one that has lost someone. If they are living, they would have to um enroll themselves um if they're cognitively capable. We have like nine different consents because there's lots of different scenarios of, for instance, I have an adult child with a rare disease um but she's not capable of doing the consent and the questions so I consented myself, provided my data, and then consented for her and then I answered her surveys.

Krista: Thank you. We have a question about whether this would be beneficial for older people with Usher syndrome like age 65 and above?

Megan: I think it's beneficial for their own whole community to get data on everyone so when people talk about a natural history study they talk about progression of disease. So if you answer a survey once, it's a snapshot. If you answer it every year it's a video, it's a, it shows the disease for that person. But you want that progression of disease shown across all ages and in order for that to be shown you need data from all ages and stages of disease.

So everybody's data is really critical um and this is an opportunity for me to say if you have a genetic report, it's really important to be able to get that data in. You upload that genetic report you don't have to de-identify. We will be manually de-identifying that into

the system. If you had one and you can't find it then contact the Usher Syndrome Coalition and they can help you go through the steps of getting access to it.

So your data is as important if not more important because you have more lived experiences than someone that's uh younger.

Krista: Thank you, Megan. Another question is "How is this platform funded and by whom?"

Megan: Excellent question. We are a non-profit uh just like the Coalition and many of the other patient organizations. So we are funded by grants, philanthropic donations and sponsorships from large pharmaceutical companies and small biotechs. That doesn't mean that those companies are getting any sort of special access to this data they have the same access that researchers from an institution would have.

The reason they are so anxious and enthusiastic about funding and sponsoring RARE-X features is that they need high quality data across many diseases and it's hard for them to do, it's not their specialty and if they do it, it doesn't go anywhere else and and so. For instance, we did not plan on having three translations in the first quarter of 2023 but it was important to pharmaceutical companies that translations move faster so they helped with that. Um so uh they have been very generous um thus far.

Krista: Thank you. I'm going to take another question from somebody's hand raised. Bill Barkeley, I'm going to allow you to ask your question and you should be able to unmute yourself and...

Bill: Yes, first thank you this is (Krista: Thank you.) uh phenomenal uh tool um I really do like the um finding out things that are linked that we may not necessarily have enough data or information on um other than some of the obvious ones. So my first question um which would follow up after I didn't get all of it just for clarity how are things like uh the like you mentioned um uh depression, anxiety and those types of measures um how are those captured? Are they separate or are they in certain parts of the questionnaire?

Megan: Currently, we have a pretty broad quality of life survey but we will have mental health surveys added as we identify them. (Bill: Okay.) You will also have a caregiver quality of life survey, we will have a burden of disease. These are all things that that will be added over time.

Bill: Okay, so yeah and some of the community conversations over time things like light in the fall um people start to experience Seasonal Affective Disorder, um establishing whether or not those things get captured obviously uh would be of help. This is probably a level three conversation but um and if you want to follow up or however you guys want to continue conversations at um on the this community has been so helpful to me in my personal journey um in understanding things, for example, to boil it down to its simple essence rather than the complexity sometimes.

On the visual side of things, it's so much about trying to understand your relation to an object -driven world and then being able to connect to accessibility, computers, technology and all that type of stuff as well as physical objects but the interesting thing on the hearing component is that a lot of people focused on questions just purely around like you know volume um and that's one thing. But then there's also discrimination. You know, could you tell vowels and consonants and can you tell what a person is saying in intelligibility. And then there's also sound and it's ruled to spatial orientation. You know, is that sound in front of me, behind me, or in back of me, and that has a lot to do with another component which is about physical safety. Am I in a safe environment or in a dangerous environment?

And then also has to do with, as my vision declines, for example, and I'm trying to read someone's face and listen to what they're saying, am I piecing up the emotional cues, whether a person is happy or angry with me. So all of those dimensions for example really play into our quality of life, play into our psychological state, and then also our physical state in terms of just how we're simply responding and reacting to the world.

So I'd really encourage and love to see the group start to be able to help that and I think that's probably the biggest power to researchers in terms of measuring quality of life and beyond. And I can't say that I'm behind that, I'm just saying this community has been tremendous in being able to bring those things forward.

Megan: That's wonderful feedback and I will tell you that a lot of those questions will be asked but they may not be asked in the same survey. (Bill: Yeah.) So we might have a fear and anxiety survey that would be available to all people in this, and we even have undiagnosed people, and um we may have um more uh hearing um specific or vision specific surveys that are really deeper dives um because we are going to have a Vision Loss Consortia and a Hearing Loss Consortia which we made up of many different diseases that have impairments and so uh there they will be making those requests as

well and there will be several uh industry partners, drug companies, that will be wanting that information as well so.

Bill: Yeah, this group has been great in helping me, for example, a lot of people and because it's all data driven, a lot of times you know people say well hearing loss, vision loss, one plus one is two. In going through this, what I see is that in our world, it's one plus one equals, you know, a factor of four. The factor of four is quality of life. When you put two, when you lose two major senses, you're losing a lot more than just the sense. Um, it starts to impact the whole body so that's why I think this is going to be a really interesting and novel experiment that could be really powerful so I applaud you guys for setting it up this way.

Megan: Thank you and the benefits will be for all patients and participants that have that situation not just Usher and that's one of the wonderful things about this is that you know we're trying to collect um as much data and share it as broadly for as long as possible and the more we have, the more analysis that can be done, the more, you know, more feedback we can get back to the communities.

Krista: Thank you so much, Bill. This is Krista speaking. That is one of the the bigger reasons that we at the Coalition are so excited about this platform as well because for years we've been talking about the need to investigate possible comorbidities, possible other symptoms that we think are unrelated to Usher syndrome but very well could be connected but then obvious the obvious things in terms of quality of life and mental health and well-being as well. So the capacity here with this this platform is so exciting because we we can, I'm just, I think it's gonna kind of unearth some information that we didn't know was there but suspected might be and that could lead to new knowledge that could lead to treatments. So um, thank you again, Bill, and I'm going to ask another question from Q&A.

Megan, can you provide any more details on the safeguards and security measures in place that our personal information and data will not be sold or shared to any third party?

Megan: It should all be covered in our privacy policy if if you are not don't see the answers to your questions please let us know and we we will provide more detail but it should be all very transparent there. We are doing this internationally, although we are still only in English, this is being built for the global community, which means that we have to go country by country and find out what their data regulations are and then we have to make sure that we are compliant. And some of you outside of the United

States may be familiar with the European GDPR compliance. It's very stringent, it's very expensive to do, it's very um complicated and we have we have met the compliance for GDPR, um but within Europe, each country has its own compliance so we take this very seriously and it's something that we are happy to answer any questions. So I would suggest that you go through the informed consent first and if that doesn't answer your questions, then you can go into the the more specifics in the privacy policy and Terms of Use.

Krista: Thank you. We have a question from a representative, a representative in Jordan. "As an NGO in the country Jordan that work with people with Usher syndrome and deafblind, can we add multiple patients or people with multiple disabilities all under one account as we will do research on Usher syndrome and different disabilities as well here." I think that that question might come up with some other partners we have in other countries as well.

Megan: At this time, and I say that because we are, you know, open to all sorts of things, we are disruptors, at this time, um a participant has to enroll themself or be enrolled by a guardian or caregiver or parent. So you could not enroll other patients. You could help patients or participants enroll. You can sit by their side, you can, um you know, sometimes a parent or a spouse may know more about your medical information than you do and so they they may want to, you might want them to join you but um, Krista, if you think that this is a request that we need to take into consideration then we will we will bring it back to our team and see how we can address it.

Krista: Thank you, a question from Megan, "Will we be informed of how our data has supported research and treatment options?"

Megan: You will be informed on a quarterly basis. We will inform the foundation how many researchers have accessed Usher data. We won't be giving out, you know, the names of those researchers because we don't want them to be hounded.

If a researcher requests that we contact a participant for a study or a trial, so the researcher would do their query and they, let's say they find 10 participants that fit their study. They would have to come to RARE-X and give us the participant ID codes because they won't know names and they would ask us to send their IRB uh um approved recruitment language via email to the participants.

Before we do that, we would say we would like you, researcher or biopharma company, to meet the organizations that are working in this disease so that they can be informed

of what you're doing and can help recruit as well. So your organization will know if researchers are recruiting people and um your organization will be told how frequently the data is being accessed.

Krista: Thank you. The question we have is, "How do I know that I have to answer a new questionnaire?" So when when we are building upon this, how...

Megan: So for instance, um we have a you know, neuro and brain survey and it asks do you have seizures but it doesn't ask how many and what kind but everybody who said yes to that question will get an email in December saying we're excited to add that we have just launched the seizure specific survey. Please log in and answer it. So if you answered yes to something or unsure and we don't have an additional survey and then we add one later you would be contacted by email. If you ignore our email and you log in when we remind you to come back every year then you would see that additional survey already added to your dashboard.

Krista: Thank you so we were planning on running this webinar until 15 minutes after the hour. We do have time for a couple more questions if there are any. I don't see any in the Q&A or the chat, um and people are able to raise their hand. I'll give, I'll give a second to see if anybody has any last questions, but in the meantime, you know, we are recording this, we will be sharing this recording with the entire community along with the ASL interpretation, the captions. You know, more information will be we'll be keep, we'll keep coming at you with more details and information that we can help clarify what this program means and um, you know, this is going to be an ongoing initiative that, you know, we'll do alongside the community. Where we are here, Nancy O'Donnell our Director of Outreach is here to support everyone who needs the support to enter their data in this into this program, into this platform, so this is not just um, this is just the first conversation so. I haven't sorry -

Megan: I just want to reiterate this isn't, we're not asking you to do a favor for a researcher or a favor for a drug company. We're asking you all to globally contribute your experience over and over again so that we can inform you back what the rest of the community is experiencing and be, inform your doctors and and the people that that are in your community um about this this um disease.

So I want you to do this for you, not as a favor to us um and think of this as part of your self-care. So you go to the doctor on a regular basis, you may have you know pharmacy um visits where you get medications, and you update this data because this is really going to be what informs the future of um Usher treatments and advocacy.

Krista: Thank you, Megan, for that, and I'll just add that you know this is definitely for the Usher community and those who are interested in advancing research this will encourage more and more and more investment in research. So we have seen the dramatic shift in biopharma investment in Usher syndrome, a rare disease, happen over the years and I do believe that's in large part because they see that there is an active and engaged community ready to be, to participate, and so the more data that RARE-X is able to capture as well to be able to show researchers this is, you know, again, de-identified data, but if anyone is looking into querying Usher syndrome or querying something else that that isn't Usher syndrome but something still shows, pops up with Usher syndrome information that's going to encourage new research, that's going to inspire new investment in treatments for Usher syndrome.

So that to me is also an incredibly exciting thing and the fact that this platform will also be capable of you know right you'll be able to upload genetic reports that is a huge request from biopharma. They need that genetic information and that is going to be, this is the potential here is, um truly exciting and we're so excited to you know continue building it alongside RARE-X and make sure it fits with what the Usher Community needs and with what researchers need, but all you know for the shared goal of supporting the community and supporting research. And the type of research is limitless.

Megan: So to our question earlier, a researcher could go in and query vision and hearing loss and quality of life. It doesn't matter what the cause of the vision loss was or the hearing loss. It is what it is, and what is the quality of life and that could then lead to recruitment of certain people to answer more specific surveys that aren't included on RARE-X so um you know, we will be marketing this freely available de-identified data to researchers of all types around the world and your organizations will be letting your researchers know about it but part of the magic of this is researchers that are not familiar with Usher stumbling across your data and um there will be lots of very interesting cross-disease things that are discovered and you know there there are some diseases that seem to be separate that may be connected and nobody's connected those dots.

So again, I don't want to keep you longer but I cannot thank you all enough, it has been a pleasure to begin to work with your community, we will continue to work with you. Um we've met several of your international partners and um we are impressed with how collaborative your community has been thus far and the fact that you all will be leading the charge with a Vision Loss Consortia is, you know, just evidence of your

commitment here and uh that there will in fact be additional surveys for your community. So again, if you have any questions, don't hesitate to reach out to us and copy Nancy.

Krista: Yes. All right, thank you so much. Thank you, Megan. Thank you to our interpreters, to Brian, our tech support. Again, yep just reach out anytime, we're here. We are going to, you know, keep this conversation going, make sure all of the questions are answered. We're adding frequently asked questions to our Data Collection page and, you know, just keep coming back and um providing that feedback. So thank you all and have a great rest of your day, evening, wherever you are. Thanks, Megan. Thank you.