>>JOHN MORRIS: Good morning everyone. We will start in just a moment. I am allowing all the attendees to come in.

Good afternoon everyone. Welcome to the International WAGR Syndrome Association Virtual Weekend. We are sorry we had a little delay, and we are hoping everyone is situated and starting to attend. We are getting our interpreters set up. We had a little issue, and I apologize for the delay.

Prior to starting our conference I want to ensure everyone can access the preferred language channel. I sent these directios a little bit ago, but those who may have missed, I will

review how to access. On your computer at the bottom on the Zoom action bar, you will see an interpretation symbol circled in red. Please select that. You will see a list of languages. Please select the best option of the languages, and I will let you know we are having an issue accessing the Italian and German. But I have the interpretation services trying to assist with that. If it is not working now please check back. And I apologize for the delay and the technical difficulties. It's what happens in a virtual world these days. If you are accessing this on a smart device, it is a little different to access the languages. You will see with Zoon more

buttons with several dots in the bottom right-hand screen. Please select that, and it will bring you to the screen where you can select language interpretation. Please select that. From there you can determine which language is your preference. Hopefully, everyone's getting settled in and we can enjoy this wonderful event together.

>> MS. MORRIS: We are the Morris family -- John and Beth and our two little girls. We are super sad we cannot see you in person again this year, but we are grateful for the opportunity to connect through technology. Today we are joined today

by 170 registrants from 29 countries. And like I mentioned, we are happy to be able to be together in any way possible and happy to share this experience with all of you. And we thank you for participating.

>>JOHN MORRIS: Again, as Beth alluded to, it's amazing how many people could come together with us although we miss seeing you together in person, but it's pretty cool to see how connected we all are. I appreciate you spending time with us today. Here is our conference agenda for today: Once our introduction is done, we will give way to Shari Krantz, our Executive Director, then go to Kelly Trout who is head of our Reseach and Medical

Advocacy. And then to our own Jenny Gunckle along with Dr. Jen Kalish who give a talk about our Registry and some really exciting news that is happening from that. And then Dr. Sue Canova from Russia will give a talk on her really exciting healthcare model as far as head of the Aniridia Centre at Research Centre of Pediatrics. And then Dr. Peter Netland of the University of Virginia will talk on glaucoma and Aniridia.

Will get started in a moment. Before that I want to go over a few things in regards to Zoom so everyone is aware. If you hover on the screen at the top or bottom, you will see the Zoom action

bar. You will see a Q&A which is an opportunity to ask questions of any of the presenters. We will do all we can to address some of those questions during the conference today. Similar to last year any questions that are not addressed during the conference we will make every attempt to complete and give appropriate responses, and we will send those both through email and through Facebook after the conference. Another option is the chat room. That is your opportunity to engage with the attendees. At the end, we will interface for one picture Right now none of the attendees can see each other, but I invite you to use the chat room to say hello to anyone. I would

ask you to please use the chat room for discussions and use the Q&A for questions. Please! Also at times like right now you will see my face. You can move the presenter box by clicking on that and sliding it on your screen. I want to make sure everyone is aware we are live streaming on Facebook right now. That is another opportunity for anyone who would like to watch it on Facebook. And we are recording today. Those recordings will be sent out via email, but we will also be saved on on our newly-designed website. Last thing before I give it to Shari: At the end please stay because I will pull everyone in as panelists and invite you to use your video feed and take a

picture. Last year it was a really tremendous experience with many people staying on and engaging with each other, and I hope we can do that again this year.

Our first speaker today will be Shari Krantz, Executive Director. She is our first staff member. In 2017 she was the first IWSA Executive Director. She has served IWSA since 2006 when her daughter, Amy, was diagnosed with Mosaic WAGR Syndrome. Shari has worked in numerous roles for the organization from a volunteer, board member, Chairperson, co-chair. And with Amy and her two grown sons, Sherry has hosted three WAGR meetings in Maryland,

meeting and supporting IWSA families around the world, and further Wilms Tumor research are two of her passions. Today she is going to provide us with an overview of some IWSA activities.
>>SHARI KRANTZ: Hi, I am Shari Krantz, Executive Director for the International WAGR Association. Here we are again on Zoom to celebrate WAGR weekend. Although we are missing the in-person meetings, it is wonderful that so many families around the world are able to be together today. We have nearly 170 people registered to participate.

For those of you who do not know, the International WAGR Syndrome Association

is a nonprofit organization governed by a Board of Directors who create the strategic plan to guide activities. Day-to-day operations are managed by me, the Executive Director, and projects around the world are carried out by volunteers. Together we represent and support 214 member families in 43 countries. The Board is a group of dedicated volunteers who meet at least monthly. Kelly Trout is one of the IWSA original founders and currently leads the Board as Chairperson. Kelly also serves as the Director of Research and Medical Advocacy and is often invited to make presentations about WAGR Syndrome at conferences and meetings around the

world.

John Morris joins Kelly and is the Board Secretary. He works closely with her on research-related projects. John and his wife, Beth, created their family foundation called Miranda's Mission which supports research and generously donates to the IWSA each year.

Nikki Hoffman is the Board Treasurer so much more. Nikki is a CPA and the IWSA's nonprofit advisor. The Board relies on Nikki's expertise to assure all funds are managed appropriately. Linda van de Sande is the first international member of the IWSA Board

and lives in Belgium. She is also the IWSA European representative and works with the Board to increase availability and access to information in support for families outside the US.

Jenny is on the Board and is passionate about the WAGR Patient Registry. She serves on the team and is interested in furthering research on WAGR Syndrome. Jenny and her family created WAGR Warriors a family foundation that support the IwSA and research related to WAGR syndrome. Jenny is the creative person behind the campaigns for WAGR Awareness Day, the Patient Registry and more.

Tom Cox has served the Board for more than a decade and was the IWSA first male Board member. Tom and his family have phosted WAGR Weekend twice. Once in Pennsylvania and most recently in South Carolina.

Rhonda Sena is a member of the Board but on the Board Emeritus which means she attends meetings but does not vote. Rhonda served the IWSA for about 20 years now in a variety of roles including as the education consultant.

One of the Board's most important jobs is to make sure that all activities follow the strategic plan. The mission is pretty simple. It's in three parts:

Awareness, research, and support. I am going to share a little bit about each of these components. But first photos like these represent the families that the IWSA supports and serves --families like yours and mine.

A few years ago the IWSA began producing an annual impact statement to provide an at-a-glance report of the year's accomplishments. Who knew that 2020 would be a year we will never forget. The worldwide pandemic affected everyone and everything as well as IWSA plans for the year. We are pleased to report that while our plans drastically changed the IWSA regrouped and achieved some great

things in 2020.

Awareness: Awareness has to do with what is WAGR Syndrome? What are the complications? Where do I go to get information? When we say promote awareness we really mean increasing knowledge of a rare disorder - knowledge for medical professionals, educators, families, and caregivers. The IWSA does this by hosting our website, attending meetings and conferences, collaborating with other rare disease organizations and researchers and clinicians who are interested in WAGR syndrome. The most important way the IWSA promotes awareness is by hosting the WAGR.org

website. The new website features translation options, is overflowing with updated information, has a section for specifically for families, and is accessible to readers and other low-vision devices. You can print most of the information and also support the IWSA by making a donation through the website. This great new tool is the result of many months of planning and writing and the efforts of numerous individuals who provided their expertise and input. We are very proud of WAGR.org. And if you have not visited the site, please try to do so soon.

On November 13 of each year the IWSA

celebrates WAGR Awareness Day. This is a great opportunity for everyone to get involved. In 2020 the theme was Make Our Future Brighter, and IWSA families around the world participated. You can see some of our smiling kids here in the photos. More than 347 individuals from 12 countries generously donated. This was the IWSA's largest fundraising event ever, and we are grateful for each and every donation. Plans are underway for WAGR Awareness Day 2021. If you are interested in joining the team to plan the event, please contact us.

Some more of our families.

Research: We know research is the key to improving the lives of individuals with WAGR Syndrome. IWSA Director of Research and Medical Advocacy, Kelly Trout, will present shortly and has some really interesting and encouraging things to share. Jenny Gunckle will also join us to talk about the WAGR Syndrome Patient Registry and why it is important part of IWSA research plans and goals.

How does IWSA facilitate research? The registry one way, but other important efforts involve developing relationships with researchers and others interested in our rare disorder. Identifying opportunities and funding

sources, providing letters of support and collaborating with the international groups help drive research forward.

Support: When the IWSA was first formed, there were a handful of families looking for information and support. Over the years the group has grown but supporting families of individuals with WAGR Syndrome has remained the top priority. This year when WAGR Weekend was virtual, 167 individuals from 25 countries came together on Zoom. This year again we have similar participation. And to make it more meaningful for non-English speaking families we provided

translation in seven languages.

Fifteen new families joined the IWSA in 2020, and each one received the IWSA's new family welcome package. In addition to a few small gifts and information about WAGR Syndrome California mom Leslie Volk and her team of volunteers continue to make quilts for each new family. Our private Facebook group now has 288 members and continues to be a safe forum for parents and caregivers to share experiences, offer and receive support, and remind each of us that we are not alone in caring for someone with a WAGR Syndrome. It's been a real lifeline for many of us over the years. Another

important way the IWSA supports families is by providing accurate and updated information on the website. There is a section created just for families. So, if you have not checked it out yet, I encourage you to do so.

More of our families.

Reporting the financial status of the organization is important and illustrates where funding came from and where it goes. Under the Board's leadership the IWSA's 2020 income was of $63,500 US dollars exceeded expenses of $59,500. Our expenses are categorized based on mission with the addition of operations. The largest

expense of the year was the new website. And as you see operations, family support and research account for the rest. With no in-person fundraising events in 2020, Awareness Day became a super important event leading the income category with almost $20,000 US dollars. The IWSA continues to be grateful for the financial support from the Saul & Theresa Esman Foundation as well as individual family and monthly donors. Facebook fundraisers are an easy way for everyone to support the IWSA, and last year totaled nearly $5,000. The IWSA received funding from the US government's PPP program and also a Delta Gamma Service for Sight grant.

The IWSA's only restricted income fund is the Amy Marshall Research Fund which was created in memory of my daughter to further research into Wilms Tumor and WAGR Syndrome. Nearly $4,000 was contributed to this fund in 2020.

More families here like yours and mine.

International Outreach: As a Board member, Tom Cox often says we need to work on the "I" in International WAGR Association. The main goal of the strategic plan is to increase international outreach. The IWSA continues to work closely with the Madoka Hawegawa in the Japan WAGR

Association. Next step has been working on appointing an IWSA European representative. The IWSA joined the European Rare Disease Organization, EURORDIS, and has been working to translate information for non-English speakers.

Transfer activities in Europe have been led now by Linda van de Sande, the IWSA's first international Board member. Linda is mom to Dylan and lives in Belgium. She has worked tirelessly to coordinate translation of information on WAGR syndrome and is promoting and encouraging enrollment in the WAGR syndrome Patient Registry. Linda collaborates with leaders and

other international organizations and is a member of the EURORDIS Patient Advocacy Group. She has had the opportunity to meet with leaders in the European Union who are involved in policy and healthcare for rare disease patients.

There are a number of people to thank for their contributions to today's event. First, the Morris family from Pennsylvania. Beth and John with daughters Miranda and Juliet planned to host WAGR Weekend in Philadelphia in 2020, but the pandemic made that impossible. The hope was to regrouop for 2021, but that of course was not possible either. So here they are

hosting Virtual WAGR Weekend for second year. Thank you Beth and John for all the planning and work that went into planning this event.

We hope you received and are enjoying your conference gift box. We mailed more than 150 boxes to IWSA families around the world. Michele Cohoon, mom to Grace and Ajay, prepared and mailed the boxes for families in the UK. Michele recently became the IWSA UK representative for new family welcome and support. She and her husband, Aaron and the children, have attended all three WAGR Weekends held in the UK. In addition to serving the Board and as our European representative, Linda

managed the conference box mailings for Europe, Russia, and Asia. Sixty boxes were prepared and mailed by Linda and her helper her son, Dylan.

In the Netherlands Jan and Chantal continue to welcome and support families into further knowledge of WAGR syndrome and its complications. They have met with researchers and clinicians at the Princess Maxima Pediatric Cancer Center, and Jan made a presentation about WAGR Syndrome and the IWSA at Grand Rounds at the University Medical Center in Utah. Jan and Chantal are parents to three children. Their family had planned to travel to Philadelphia for WAGR Weekend

in 2020 and then again this year in 2021. Both trips had to be cancelled, but we hope another trip can be planned in the near future.

Many thanks to our friend, Galina Gening. Galina works closely with the IWSA to support families in Russia. She is president of the Interregional Support Center for patients with Aniridia Syndrome and WAGR Syndrome in Moscow and is a trustee to Aniridia Europe. Galina is an amazing advocate for individuals with Aniridia Syndrome and WAGR Syndrome. Kelly Trout and I have had the pleasure of meeting in person with Galina and several Russian ophthalmologists. Linda is also

working closely with Galina to encourage Russian participation in the WAGR Syndrome Patient Registry. Thank you for all you do, Galina, and we look forward to continuing our work together in the future.

Over the years the IWSA is dependent on the dedication of volunteers who bring special talents, skills, and perspectives to the organization. Some people serve on the Board or host a WAGR Weekend event, while others write stories for the website or translate information. Many families have hosted successful fundraising events or helped with WAGR Awareness Day planning. If you have an interest in contributing to

the IWSA and supporting the families, please contact me a Board member or email us at reachingout at WAGR.org. If you have questions about the IWSA or something you would like to share, or if you want to get involved with the organization, please contact us. Thanks for the opportunity to share my presentation today, and I hope you enjoy the rest of the speakers.
>>JOHN MORRIS: Thank you so much, Shari, for the organizational updates. Your efforts in leading the organization are greatly appreciated.

I do want to point out that this year we have interpreters. The organization continues to try to improve access.

Unfortunately, this year we were unable to provide a live Norwegian interpreter. But I do want to point out that we have closed captioning services available for people who prefer to read Norwegian. Within the chat room, Danielle, one of our support folks has put in a link to StreamtText for anyone who would prefer closed captioning for a Norwegian language. You can please access that now. Look for that. If you scroll up, you will see it. At the bottom of the screen are the interpretation channels.

Please mute the microphones.

Our next speaker is Kelly Trout. Kelly

is the co-founding member of the IWSA, and is currently serving as the Chairperson of the IWSA Board of Directors. She is currently serving as the Director of Research and Medical Advocacy. Kelly assists families and physicians with questions about diagnoses and treatment of WAGR Syndrome and related disorders. She is also the Program Manager for the IWSA Patient Registry. She's written numerous articles on WAGR syndrome for many publications. Today she is going to be discussing some of the exciting WAGR
Syndrome Research being done around the world.
>>KELLY TROUT: Hi, I am Kelly Trout,

and I would like to add my welcome to Virtual WAGR Weekend 2021, especially to those folks with us for the very first time ever. I wish we were meeting in-person. But for now it is wonderful to know we are all watching this together from all over the world. Today I am going to talk about research on WAGR Syndrome And telling you about some very exciting and hopeful projects that will help our kids to live better, healthier lives. I will tell you a little bit how the IWSA helps make research happen. And I will talk about how you and your child can be a part of these efforts.

First, I want to start out with this

picture of my daughter, Caroline. I can hardly believe it, but next week Caroline will be 40 years old. Forty years ago there was no such diagnosis as WAGR Syndrome. There is no name for it back then. It was just called Aniridia and Wilms Tumor. When we got this diagnosis, I tried to find all the information I could about it. After weeks of searching, I found one paragraph in one medical textbook about one patient. And that was it! There simply was not any knowledge or information about this disorder, and there certainly was no research going on.

I remember feeling desperate and angry.

How could I help my child if doctors did not know anything about WAGR Syndrome? When Caroline was a teenager, the Internet came along, and it changed everything. It allowed parents of kids with this very rare disorder to find each other and to communicate. By communicating, we discovered that our children had a lot of similar conditions. Similarities that even doctors did not know about. We began to understand that if doctors were ever going to learn about WAGR Syndrome, we parents and families were going to have to be the ones to teach them.

Now while we would love to have a cure

for WAGR Syndrome, We know that is not possible yet. But a lot of things are possible. The fact is there is still a lot we do not know about this disorder. It would be a huge help to simply understand the syndrome better. For example we now know that WAGR syndrome involves a lot more than just the acronym of Wilms Tumor, Aniridia, genital urinary abnormalities, and a range of developmental delays. There are many other conditions that are possible like chronic kidney disease, seizure disorder, behavior problems, and many others. We need to identify all of these conditions and determine how often they occur, and why they are related to WAGR Syndrome so that

parents and doctors will know what to look for and how to find these things early so they have the best chance of being treated effectively.

We also know that Wilms Tumor in kids with WAGR Syndrome is different in some ways than it is in typical kids. There is growing evidence that Aniridia may be different too. We need to know a lot about these differences in order to get the best treatment. Developing standardized guidelines for care would also be an enormous help. Have you ever taken your child to the doctor and realized they don't really know what to do? Having standards of care would be all the difference in the world. So

how can we get standards for care? Well, you can't just make them up. You have to collect data, and collecting data is research.

So research can give us all the answers our children need. But medical research is very expensive. So how can a very small organization like the IWSA make research happen? Well, it turns out that money is not the only thing it takes. Research requires gathering data on patients such as through a patient registry. It takes making connections with researchers and developing relationships with them. It takes the resources of time and effort of volunteers, and it takes sharing

results with parents, with doctors, and with other researchers around the world. It turns out if you have all of these things, getting the money for research, well it is not easy. But it does become much more doable. So tthe IWSA focuses on doing each of these things.

First the patient registry: Many of you already know about the WAGR Patient Registry because your child is already in it. If that is the case, bravo! If you do not know about the Patient Registry, our own Jenny Gunckle will talk about it in just a few minutes and fill you in. But in terms of getting research done on WAGR Syndrome, The

most important thing to know is that enrolling your child in the WAGR Syndrome Patient Registry is the single most important thing you can do. So please listen carefully when Jenny comes on.

Creating and building relationships with scientists is a critical part of getting research done. The IWSA works hard to find scientists who are working on Wilms Tumor, on Anirida, and all the other conditions related to WAGR Syndrome to get them interested in researching WAGR Syndrome and finding new ways to help our kids. Parents and families that are part of IWSA are not just the heart beat of this

organization. They are the engine that drives it. Each of us can do something from fundraising to raising awareness in all sorts of different ways to supporting other families by volunteering your time and skills. All of these things work together, and all of them are absolutely vital to making research happen.

Another important part of research is sharing the results. We want the world to know what is happening in the world of WAGR research. So we share the results on our website, on printed materials you can share with your doctor, and through presentations to scientific conferences. So put all

these things together: The patient registry, relationships with researchers, WAGR families volunteering their time and efforts, and all of us sharing data and information. What you get? Well, here is Dr. Vicki Huff, Dr. Cristy Ruteshouser, and mouse lab director, Nikki Williams dressed up in their PPE on the way to work on the Wilms Tumor mouse project. They are testing a medication that could actually help prevent Wilms Tumor in kids with WAGR syndrome. With COVID-19 this past year, things have been really challenging. But I am happy to tell you that this project is back on track now and is making excellent progress.

Here we have Janna Hol who recently published a study that compared children with WAGR Syndrome and Wilms Tumor to typical children who develop Wilms. Dr. Hol and her colleagues stressed the importance of our children being followed at pediatric cancer predisposition clinics when possible.

Many of you are familiar with Dr. Jeff Doan who published several times in the past year including a textbook called Management of Genetic Syndromes, a book that is considered a go to guide for many pediatricians.

Doctor Andrew Murphy is another

physcian who has developed a keen interest in WAGR Syndrome. He is working on a project that could lead be to ways to use genetic testing to predict which children with WAGR Syndrome Will develop Wilms Tumor.

Yet another familiar face is Dr. Joan Hahn. Many of us remember Dr. Hahn from her study of WAGR Syndrome at the National Institutes of Health in 2006-2014. Dr. Hahn is still very involved with studying WAGR. One of her recent projects involved a study of whether gene editing could fix some of the brain abnormalities our children have. This project is called Basic Science. And it is a long way from

basic science to having an actual treatment. But the theory proved to be correct. And that is amazing. Just like the very cool zebra print glasses she is showing off here.

For several years now Neil Lagali and his colleagues in the organization Aniridia Europe have been accomplishing remarkable things in terms of learning more about Aniridia and its complications. Unfortunately, one thing they are learning is that problems with the cornea and be worse in patients with WAGR Syndrome than in patients who have isolated Aniridia. While that it is disappointing, it is really important information. It means

that parents and doctors need to be especially careful about our children's corneas. Aniridia Europe has created wonderful videos about how to do this. And you can find them on the WAGR.org website.

Here is Dr. Jen Kalish. You are going to see her in a few minutes. Dr. Kalish is a geneticist at Children's Hospital of Philadelphia. Thanks to our WAGR Weekend host, Jenna Beth Morris, we were able to interest Dr. Kalish and her team to take a look at the data in the WAGR Syndrome Patient Registry. What followed has become a full-fledged and ongoing collaboration between members of the Kalish lab and

the IWSA Patient Registry team. I am thrilled to tell you that we have just submitted an article on the registry data, and I can tell you it will be huge. It will be the first publication to recommend a model for managing medical care in WAGR Syndrome. All of it is based on the evidence provided by participants in the Registry. We will have lots more information about this article soon.

But for the moment I hope all of you take great pride in knowing that you did this. By participating in the registry you changed the world for your child. And you have changed the future for all of our children.

Speaking of the future, I want to give you an update on the clinical Center of Excellence for
WAGR Syndrome. You may remember we started working on this project a couple of years ago when Children's National Hospital announced plans to create a are disease institute that would house clinics for rare disorders like WAGR. The beauty of a center of excellence is it can become a one stop shop for all the specialists a child with WAGR syndrome might need. On top of that, over time the specialists at such a clinic become experts in WAGR Syndrome. Can you imagine taking your child to such a place? Well COVID-19

slowed things down, but it has not stopped it. I am happy to tell you that construction of the Rare Disease Institute is now complete. And we hope the center will be able to start seeing WAGR patients as early as this winter. Look for lots more information about this in the coming months.

Here's Caroline again. So much has happened since she was a baby. From no name for her diagnosis and almost zero information about it to a broad spectrum and an explosion of research information. From no help to real hope. I will leave you with this: Consider your own child's journey. They are standing on hope, too. What

will you do to help them get there? Thank you!

>>JOHN MORRIS: Thank you so much, Kelly. I always look forward to these updates. I do want to remind people that we do have the Q&A so as presenters provide these wonderful and valuable experiences, I imagine some questions will arise. Please feel free to post those. We will do our best to address some of those questions. And, if we cannot do it today in a timely fashion, we promise we will answer those in a brief timeline.

Our next discussion will involve the IWSA's Jenny Gunckle and Dr. Jen Kalish. Jenny Gunckle joined the IWSA

Board of Directors in 2017 and is also a member of the IWSA Patient Registry team. Jenny lives in Michigan with her husband, Adam, and her children Emma with WAGR Syndrome and Evan. She has a passion for furthering research specific to WAGR Syndrome and for encouraging participation in the IWSA registry. Many of you will recognize her from the registry campaign. Dr. Jen Kalish is a clinical geneticist and physician scientist at Children's Hospital in Philadelphia and the Perelman School of Medicine at the University of Pennsylvania. Her focuses on Beckwith Wiedemann Syndrome and disorders of growth and genetic cancer predisposition. BWS is similar

to WAGR sydrome as it is a rare condition where patients are predisposed to developing Wilms tumor. Today her and Jenny will share some of the results of the registry campaign and how valuable this informatio is to reserachers and how it is being used.
>>JENNY GUNCKLEHIM: Hi, everyone. My name is Jenny Gunckle, and I am an IWSA Board member and mom to Emma who is 8 years old. I am here today to provide all of you with a really exciting update on the WAGR Syndrome Patient Registry. Since the registry was developed by the IWSA in 2015, we have said over and over again this data provides hope for those living with WAGR Syndrome, and I hope you will

understand a little bit more about what this means after you hear more about what has been happening over the past year.

Before we get to the good stuff, for those of you that might not know, the Registry is an online questionnaire of 77 questions in an IRB approved survey hosted on a secure server by the Coordination of Rare Diseases at Stanford (CORDS). It can be completed online or with printed copies by adult patients themselves or parents or guardians.

Currently we have over 140 patients with WAGR Syndrome represented in this

data. It means our registry has the largest collection of standardized information about individuals with WAGR Syndrome In the world. The main purpose of the registry, of course, is for our data to help better understand the conditions associated with WAGR Syndrome and to potentially develop treatments or therapies that could help improve the lives of our kids and future kids to come. As with WAGR Syndrome, when so few patients with this disorder exist, getting each and every patient around the world to enroll in the registry is so important. As many of you might remember, last summer we launched a full-scale campaign to increase enrollment in our

Registry. You might have seen one of our many videos. We actually heard from over 50 people talking about the benefits of enrolling and annually updating your registries. If you missed any of these videos, you are able to find them anytime on the IWSA public YouTube channel.

At the end of the day, this campaign exceeded all of our wildest expectations. It increased our records by almost 80 percent. So to each and every one of you who took the time to enroll or update your registries, thank you. Honestly, whatever amount of time it took you to do this, was well worth it. This is because in the past year,

we have had three researchers obtain approval to access our Registry data. A few fireworks! [Laughs] These include a geneticist who is studying the gene mutations associated with Aniridia, a team of pediatric cancer researchers who are studying Wilms Tumor and WAGR Syndrome, and a team from the Children's Hospital of Philadelphia led by Dr. Jen Kalish. Dr. Kalish spoke at last year's WAGR Weekend, and she is here with us today to provide an update on the unbelievable progress her team has made this year and some of her exciting plans for the future.

>>DR. JEN KALISH: Hello, I am Dr. Jennifer Kalish, a clinical geneticist

and researcher at Children's Hospital of Philadelphia. My clinical work and research focuses on studying cancer predispositon syndrome such as WAGR Syndrome and Beckwith-Wiedemann Syndrome. This work includes clinical and characterization of these rare syndromes and developing clinical care guidelines. Registries are essential to understand how to diagnose and care for patients that have rare diseases like WAGR Syndrome. The data collected from Patient Registries helps us to learn about the experience of each patient and what medical issues they may face. We use then information to guide physicians and healthcare providers to try to improve care for

patients with WAGR Syndrome.

In order to study patients we need to systematically collect patient data in the patient registries like to WAGR Syndrome Patient Registry. The more patients that are available to study the more powerful the collection of data becomes. A challenge in rare disease research includes the small number of patients affected by a given condition. This means with rare diseases like WAGR syndrome every patient who participates in the Registry is very important. And we need as many patients as possible to join. The information for those who join the WAGR Syndrome Patient Registry

already has helped us to understand more about the different features affecting patients. Your information has shown us that WAGR Syndrome has a number of common features beyond the classic definition of the syndrome. Therefore, in collaboration with the International WAGR Syndrome Association we now consider WAGR a spectrum rather than just a syndrome.

We recently submitted the summary of findings for publication in a medical journal with the goal to include the characterization of the clinical features in the WAGR spectrum. We have developed updated clinical care guidelines for patients and families

with WAGR specturm to share with their own care teams. We anticipate these guidelines will extend the current approach to WAGR spectrum. In addition we have submitted abstracts to several genetics oncology conferences to share this information with the broader clinical and scientific community. We are also working on developing additional projects and questions and are always interested always in learning more about family-centered research priorities.

For those of you who have already joined the WAGR Syndrome Patient Registry, thank you as this project would not have been possible without

your participation. You are welcome to update your information anytime to help us learn more about the long-term outcomes of WAGR spectrum at any time. For those that have not yet joined, please consider joining because the more data that is shared the more we can work to improve understanding and care for WAGR spectrum. We look forward to sharing more data from the work I have just discussed in the coming months, and I thank you very much for your attention today.
>>JENNY GUNCKLEHIM: Dr. Kalish and her team have invested counless hours collecting and intepreting our data. And honestly what is happening righ now is a dream come true. Over the next

few months you will be hearing a lot more from us and from Dr. Kalish talking more specifically about this article and how it can benefit your child's care. So please be on the lookout for all of that information coming soon.

As Dr. Kalish and others continue to move forward with the research into WAGR Syndrome, there really is no better time than now to enroll or update your registry data. And you can find all of the information you need to do that on our website at Wagr.org\Wagr-syndrome-patient-registry. Hopefully, you can see that on your screen.

You can also find the Registry page from our Wegr.org website. Just navigate to wagr.org and our homepage. You can see the tabs at the top. If you go under research tab there is a link there - wagr syndrom patient registry. Click that, and it will take you cirectly to the page. At the top there is an orange button that says enroll or update now. It will take you to that CORDS enroll page where you can enter your login information if you created that, or you can click not yet a participant, and you can sign up to enroll in a new registry.

Going back to IWSA page there is a lot

of informatio on this page you can go through on your own. The top video is a Youtube video that was a video created by patients with WAGR Syndrome. If you have seen this, check it out. It is worth the couople minutes to watch.

Another thing to point out: If you go down further on the page there is a question that says: How do I register? Right here is a registry FAQ with a bunch of questions and answers on how to enroll on the registry and common questions. You can click to get English or in multiple other languages. Just click the orange button to find it in other languages.

Also, under Why should I register, it will take you to the YouTube channel. If you click it, it will take you to YouTube where you will see different available videos on the channel. If you go down a bit, you will find videos for the WAGR Syndrome Registry campaign last summer. There are also other videos.

If you have questions on the registry, I highly recommend taking a peak at the webpage. As I said before, with such a small patient population like ours, every single entry makes a really big difference. So please reach out to us if you have any questions or issues with enrolling. If there are any

barriers to enrolling online, please let us know, and we can mail you a paper copy. The Registry is in English, but if it challenging for you to complete it, please feel free to reach out to someone you trust - a friend, family member or healthcare professional or your country's Aniridia organization to help you with the translation.

With this registry data comes power. The power to know whether something is a WAGR thing, the power to formally publish care guidelines to help our kids get the treatments they need, and the power to help entice researchers to further study our rare disease.

>>EMMA AND JENNY: Hope is one short survey away. Thank you everybody so much, and we hope to see you again in the future years in person WAGR Weekends. Have a great day everybody. Goodbye.

>>JOHN MORRIS: Thank you so much, Jenny and Dr. Kalish. Please keep up the good work. Jenny, I hope Emma knows now she is famous. Personally, I am so excited to see all these research projects proceed and what additional opportunities may arise.

>>MORRIS CHILDREN: Hi.

>>JOHN MORRIS: I also hope everyone is ready to update your Registry after this presentation. I did see within the chat room that a link was added

within the chat box. Jenny shared it during the presentation but please go to the chat room for the link. I see a really great question about how often we should update the registry. Kelly Trout indicated whenever your child has a new diagnosis and at least once a year. Thank you, Vera, for the great question and thank you, Kelly, for answering it.

Before we keep going, I want to give a quick look at our agenda to see where we are. Our next presenter is a Dr. Cannova, a research and clinical ophthalmologist and head of the Center for Anirida Research Institute of Pediatrics and Children's Health at the

Central Hospital at the Russian Academy of Sciences in Moscow who will talk about the services provided at the Center for Aniridia research. This includes patients with WAGR Syndrome. The center is a model of the kind of medical care that the IWSA believes would be beneficial for all patients with WAGR Syndrome. As Kelly mentioned earlier, IWSA is working to launch a similar center in the US soon. We are launching a Center of Excellence soon, and we want to encourage the development of clinical centers of excellence for patients with WAGR Syndrome around the world. Here is Dr. Cannova to tell us about this innovative and exciting clinic in

Russia.

>> DR. CANNOVA: Greetings to all listeners. I am the head of the Aniridia Center. I would like to thank the organizers for the invitation and opportunity to tell you about the Russian Center for Aniridia. My report has a short presentation of the department and opportunities patients have when they come to our center.

[READING SLIDE] We perform research and real-world patient management with the goal to tackle urgent problems of preventive pediatric medicine in public healthcare. Our center is a modern comprehensive hospital infrastructure designed for maximum care. We offer a

full range of clinical ophthalmologist services. We treat and provide diseases of the cornea, and diseases of the retina and macular
[READING SLIDE]

Patients from all over Russia come to get advice.
[READING SLIDE]
Multidisciplinary approach ensures prompt diagnosis treatment and rehabilitation of a variety of diseases in adults and children 0-18 years of age.
{READING SLIDE]
>>JOHN MORRIS: Thank you, Dr. Cannova. Your clinic is an amazing medical model, and thank you so much for

sharing this with our families.

Our next speaker is Dr. Peter Netland. Dr. Peter Netland is the Scott Mosten Professor and Chair of the Department of Ophthalmology at the University of Virginia School of Medicine in Charlottesville, Virginia. In addition to his recognized expertise in the clinical management and surgical treatment of glaucoma, Dr. Netland is an innovative and prolific investigator. He has written more than 300 perr-review publications and published six textbooks. His interest in amiridia has led him to serve on the boards of several aniridia nonprofit organizations. We are fortunate to

have him with us to speak about glaucoma.

>>Dr. NETLAND: I would like to thank the organizers of the WAGR Weekend for this very nice invitation. I really appreciate it; I would also like to welcome all the participants from all over the world.

My name is Peter Netland. I am with the University of Virginia, and I will be talking about Aniridia and glaucoma. Glaucoma is potentially a vision threatening problem that is the cause of irreversible vision loss. It is common in Aniridia and any age can be affected. Oftentimes it occurs in childhood in aniridic patients. The

treatments for glaucoma are effective so it is important to try to identify this disease while it is asymptomatic so it can be treated before any vision loss occurs.

There are various causes of aniridic glaucoma in terms of the mechanisms. In newborns infantile glaucoma is fairly uncommon in aniridia. It is usually due to some sort of coexisting anatomical problem. More commonly this occurs a little later in childhood and early adulthood. The most common form is an open angle glaucoma. Angle closure has been described especially after other surgeries and a progressive angle closure is also described but

this probably relatively uncommon.

We did look at the mechanisms in some detail in a comparative study. Out of 86 eyes, 27 patients were diagnosed with glaucoma and 16 had no evidence of glaucoma. [READING SLIDE] In this group anirida was the most common diagnosis. There were no WAGR patients in this group.

The findings are shown here. The majority of the patient had open angles. [READING SLIDE]

This is a typical findings on the study. [READING SLIDE] this was a closed angle. This is a photograph of

an eye with Aniridia Syndrome glaucoma. [READING SLIDE]

Glaucoma is fairly common. 46% rate of glaucoma. The age of diagnosis, the mean was 13.6 years in the median was 8.5 years. So about half patients were diagnosed with glaucoma by 8-1/2 years of age. Most of patients with glaucoma require treatment with medications and the majority eventually require some sort of glaucoma surgery. We measure pressure in these patients to help understand if patients are developoing glaucoma. Increase usually in pressure. The gold standard is applanation tonometry. Rebound tonometry we have found useful as well. This does not

require an anesthetic and can be used in young children without having an anesthetic. Corneal thickness is an influencer which is increased in anirida. We take that into account when interpreting measurements to understand if the pressure really is elevated.

[READING SLIDE]

We recommend examinations about every six months for patients who have not developed glaucoma. Will increase if patient develops glaucoma. Age is a risk factor.

The treatment for anirida is effective. Laser is not very useful. Type of survery varies.

[READING SLIDE]

Drainage implants are sometimes used.
[READING SLIDE]

We are interested in new minimally invasive glaucoma surgeries. They are used frequently in adults less information about these procedures in children.
[READING SLIDE]
These are some examples of treatments that have been used and are promising. We do need to collect some information about these before we can recommend these.

In conclusion, aniridia is often

associated with glaucoma causing progressive vision loss. Patients often require surgical treatments which generally have good outcomes.
[READING SLIDE]
Prompt treatment and monitoring may prevent vision loss due to glaucoma and aniridia. New treatments are promising.

I would like thank IWSA for putting together this great conference. I would also like to thank Aniridia North America - NA, Vision for Tomorrow and Aniridia Europe. I especially want to thank the patients I am following and the families and my collaborators who I am very, very appreciative for and very

appreciative to. Thank you all very much for listening in, and I hope the rest of your weekend is informative, productive, and enjoyable. Thank you very much. >>JOHN MORRIS: Thank you, Dr. Netland. And speaking for our family, we can't thank you enough for your ongoing efforts within this field. So we are done with the presentations, but I really hope everyone will hang out for a couple minutes so I can go through a few things and then we can get that cool picture of us together. So I hope everyone has enjoyed these talks and hopefully many of you have had your questions answered. We will look through the list of questions. And, as we did last year, we promise to

give accurate and appropriate responses to questions not answered today so look out for an email with those responses.

For those not able to participate last year, we have a wonderful redesigned website. I encourage you to visit it for information pertaining to support, awareness, and research. We will also host recordings of this event for future viewing purposes. Please be on the lookout for information later this year regarding new journal publications and the study in our registry data. This publication will better educate the medical community on the many features of WAGR to entice researchers to take the next step in studying.

This is really big and amazing news. And once published the IWSA will share a lot more information in how you can use this information. Of course none of this would have possible for each of you taking the time to enroll in the WAGR Patient Registry. With such a rare disease every entry from around the world makes a huge impact. If you have not enrolled, there's no better time than now. If you have not updated your information this year, now is a great time to do it. You can find the link to the Patient Registry on WAGR.org. If you have any questions or issues, please reach out. It is amazing how much power can come from a 77-question survey which you can fill

out any time from the comfort of your home.

Again, thank you everyone. As a reminder: At the end you will see a pop-up on your computer to complete a survey. We appreciate that. But, if you missed that opportunity, it will be emailed to you tomorrow. The videos will be available to you shortly. You can visit WAGR.org to find those. Again, will get the Q&A out.

I hope to see as many of you as possible for WAGR Weekend 2020. Hopefully, we will be together physically. What I would like to do now is I want to get everyone on the

same screen. I will invite everyone now to be a panelist and when you get the invite, please accept it and please turn your video on. Give me a minute. You also want to change it to a gallery view.

Please turn on your video as you come on.