



WINGS

WAGR/11p Deletion Information, News,
Gorilla Stories

International WAGR Syndrome Association
P.O. Box 392 Allen Park, MI 48101

www.wagr.org
Spring/Summer 2011

*To all our IWSA families,
friends and donors,*

Tremendous progress has been made during the past several years in many areas of medicine and medical research. One of the most exciting and productive areas is the field of genetics. Identifying and pinpointing specific genetic abnormalities and deletions is already helping to understand the causes of diseases and syndromes, including WAGR Syndrome. The short arm of the 11th Chromosome (11p) is the affected area for WAGR Syndrome.

According to Dr. Joan Han, MD, and lead researcher for the WAGR Syndrome/11p Deletion/Aniridia studies at the National Institutes of Health, "WAGR is classically thought of as having PAX6 and WT1 deletion, but 11p Deletion is a more encompassing diagnosis because it would include not just individuals with PAX6 and/or WT1 deleted, but also those for whom any chromosome 11 genes are deleted."

The International WAGR Syndrome Association is announcing the name modification--WAGR/11p Deletion Syndrome--to IWSA and WAGR families, other nonprofits and support groups, and medical, research and press organizations. This modification is necessary to more accurately reflect the actual genetic

description of the syndrome. We encourage all of our WAGR families to use this newly updated name--WAGR/11p Deletion Syndrome--effective immediately. To help with this change and to maintain consistency, please join us in using the "new" name, noting the specific capitalization and spacing. We feel strongly that this change will help to better clarify the genetic nature of WAGR Syndrome and hope you will all join us in making the change a swift and seamless process.

If you have questions or comments, please contact IWSA President, Catherine Luis at catherineluis@msn.com or IWSA Health Consultant and Board Member, Kelly Trout at kellytrout@sbcglobal.net

FLOWER POWER
FUNDRAISING



Think Spring and help the IWSA! By participating in our FlowerPower fundraiser you can help us reach our goal of \$1,000. Fifty percent of all sales go to the IWSA!

Visit the following website
<http://www.flowerpowerfundraising.com/it/73141/3e5JZ889j091>

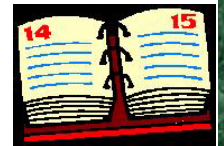
Thank you--your purchases will help our organization to keep blooming!

Order deadline is April 30, 2011

WAGR Get Together Planned for the UK

by Jenny Langley

The date has been set for the annual Aniridia Network UK meeting. It will be held on **Saturday, May 14, 2011** in Birmingham (which is located in the middle of England).



We are planning to go to an activity centre, Ackers Adventure and plan to have speakers in the afternoon.

We hope that WAGR families residing in England or close to the vicinity will plan to attend. A buffet lunch is also planned.

For more information or to RSVP for this event, please contact Jenny Langley at jenny.langley@aniridia.org.uk

We hope to see you there.

On the Inside

Gorilla Stories	pg 2-6
Cerys' visit to the NIH	pg 6-8
NIH Update	pg 8
Rare Disease Day	pg 8-9
Newly Formed Committee	pg 9,11
Hockey and Hope II	pg 10
WAGR Weekend 2011	pg 11



Our 'Ray' of Sunshine

In November of 2008, my husband James and I along with our daughter Sydona were thrilled to welcome a new baby girl into our family. I had a great pregnancy with no complications. Rayleigh Jean was perfectly beautiful, we were so thankful for a healthy baby. While still in the hospital we noticed her eyes were swollen which seemed strange since she was born via c-section. She also didn't open them much, so we questioned the doctors but they reassured us it was normal and everything was fine. We let it go and headed home as a family of four.

A few weeks later we began to wonder if something was wrong with her eyes. They were so dark and she wouldn't focus well on anything. We talked about it a little but I kept telling myself; *the doctors have seen her several times, they would have noticed if something was wrong, or she goes for a check up at two months, if we're still concerned, I will ask then.* I did NOT want to believe that my baby could have something wrong. Then at six weeks old I was sitting with Rayleigh, talking to her face to face trying to catch a glimpse of her looking into my eyes. And then it hit me

and I thought, *My baby can't even see me! My baby is blind!* I still didn't call the doctor or anyone for that matter; I wasn't ready to voice my fears out loud. I just cried and prayed. I left Rayleigh with my sister that day and went to a doctor appointment for myself. When I got out of the doctor my sister called and said "I really think something is wrong with Rayleigh's eyes, don't you think you should get her checked out?" I knew then I had to bring her in. We got an appointment right away and thankfully my dad came with me since James couldn't get off work. I was so nervous as the doctor looked into her eyes and then felt as if my knees might give out when he said, "It looks like she doesn't have an iris, and I've actually never seen that before." *What???! He's a doctor and he's never seen this? This is bad! He is supposed to have some simple explanation and say everything will be fine!* So many things were going through my head. *What does this mean? Can she see? We'll never know her eye color? They can't fix this! How am I going to tell James?!* I managed to keep it together probably out of sheer shock, as the doctor set up an appointment with an ophthalmologist. As soon as we left I started searching 'no iris' on my phone and I found Aniridia. As I read about it I could hardly believe it, I knew she hadn't been diagnosed yet, but I knew it was what she had. *How can this be happening?* I read words like cataracts (*babies can have cataracts?*) nystagmus, glaucoma, then Wilms' tumor, and WAGR Syndrome. It was all too much to take in but yet we had to. After a very LONG weekend we went to her appointment. The doctor did an extensive exam and then confirmed our fears. She told us Rayleigh had

aniridia, nystagmus, and that she also had cataracts in both eyes that needed to be removed right away. She mentioned WAGR Syndrome and said Rayleigh needed genetic testing to see if she had that as well. She set up an ultrasound for her kidneys to look for signs of Wilms' tumor and a consultation for her cataract surgery. I will never forget that day, hearing all this about my precious baby that was now sleeping peacefully in my arms and looking across the room at my husband with tears running down his face. The best word to describe it all is 'devastating'. Our dreams of having a healthy second baby had been drastically changed in that moment.

We went right into the mode of doing whatever we had to do for her. She had her cataracts removed and implant lenses placed in her eyes at three months old. We then had to do more testing to find out if she had the WAGR Syndrome deletion. It was crazy how we went from hoping and praying nothing was wrong with her eyes to, hoping and praying it was ONLY her eyes. We weren't expecting any results for about two weeks so when they called four days later, it was again shock and another hard blow as she told me the tests showed Rayleigh had WAGR Syndrome. It felt like a very bad dream, I cried more in those few months than I ever had in my entire life. I felt guilt, anger, sadness and fear. There were so many different emotions at once. But one thing we're learning on this journey, is that God often uses the most difficult things we face in ways we never could have imagined. This is definitely a growing process...we still have our hard days. The days when I wish she didn't have to struggle to reach that milestone that comes so easily to most. Or the

days when I feel envious of moms bringing their kids for a simple checkup, while I bring mine to see if cancer has started to grow on her kidneys or if the pressure in her eyes is going to cause her to lose more vision.

Thankfully, Rayleigh's health and medical issues have not been too serious thus far. Her vision is quite good, you may not even realize she has a vision impairment, unless you see her stop at the edge of the carpet in an unfamiliar place unsure if it's a step or not. She has no glaucoma and has had no issues with her implanted lenses. She wears her glasses well and they do help to somewhat improve her vision. We did find out after a routine ultrasound last August that she has Nephrogenic Rests on both kidneys. We continue to monitor them closely for any signs of change or growth in which case we would have to look into treating them due to the risk of them becoming Wilms' tumor. Rayleigh has had sleep issues from the beginning, so up until she was 22 months old we were up with her every night, sometimes hours at a time. She sees a sleep specialist and after failed attempts of trying different techniques without medication, she now takes Melatonin to get her to sleep at a good time along with a medication that helps her stay asleep through the night. Hopefully one day she will no longer need this, but as for now we are thankful that we are all getting better



THE LAJOIES ~ 2010

rest. Other than the things we continue to watch for, Rayleigh has been a healthy girl. She sees a Vision therapist twice a month, an Occupational therapist for some 'fine motor delay', and a Speech therapist for 'mild expressive speech delay' once a week. She did some Physical therapy before she learned to walk but was released since they felt any physical milestones she reaches a little late are due to her vision impairment. She is doing great with her therapy and continues to surprise us as she progresses at her own pace. Her speech has picked up a lot in the last few months, which is so exciting as she is able to communicate her needs/wants to us much more easily.

Rayleigh LOVES her sister Sydona, who she calls "Noni". Its been amazing to see how wonderful of a big sister Sydona is to her. She has always been Rayleigh's #1 fan (well after me of course), she is so proud of every little thing Rayleigh does. It warms my heart when she randomly says "Oh my word, she is SO cute!!", like it's the first time she's ever seen her. I know God had hand-picked her to be there for Rayleigh, to teach her, to protect her, and love her, long before Rayleigh was even born. She makes us proud!

Rayleigh today is a happy, loving, funny two year old girl that touches the heart of everyone she meets. She has taught us and everyone around her so much. I've learned not to take things for granted, and to celebrate the little things in life. I'm learning to live in the moment, its not always easy but I try not to worry about what the future may hold. We are so thankful to have come to know, even if its only through Facebook and Email, such amazing and inspiring

families that are on this WAGR journey alongside us. We have been blessed to be surrounded by wonderful, supportive family and friends, but to also have those that can truly understand what it is to have a child with WAGR/11p Deletion Syndrome is a huge comfort. It is amazing to look back and see how far we've come. And see the encouraging words like "It will get easier" or "You will adjust to your *new normal*" actually becoming reality. There were certainly times when I felt it was all too much to bare. But we have seen the faithfulness of God; He has lead us through every trial and I know He will continue to do so, no matter what may come. As someone once said "If God brings you to it, He'll bring you through it." We choose to trust Him.

Over the past two years we have seen the confusion and questions of it all change from, "Why did this happen to us?!" to "What a blessing and privilege it is to be the parents of such a special girl!" And we wouldn't have it any other way. Like I've said since she was born she is our '*Ray*' of *Sunshine* and she truly SHINES!!

Written by Destiny Lajoie, SC

IWSA Officers & Members

President - Catherine Luis

Vice President -

Tammie Hefty

Secretary - Julie Dell

Treasurer - Jeffrey Hefty

Gift Coordinator -

Elizabeth Duffy

Professional Advisor -

Nikki Hoffman

Board Chairperson -

Annie Prusakiewicz

Member - Shari Krantz

Member - Rhonda Sena

Member - Kelly Trout

Hope and Acceptance

Our beautiful little girl, Audrey Grace, was born 15 months ago. It has been a full journey, with many ups and downs, but beyond all the diagnosis, our little girl is the love of our lives and continues to thrive and grow despite the challenges she faces.

When Audrey arrived into our world, Harry and I were feeling very confident that we would have a healthy baby. We were the pictures of good health, hardly ever visiting doctors throughout our lives. The pregnancy was uneventful. We had no doubt our baby would be "perfect." The morning after Audrey was born; doctors turned our lives upside down. At first, it sounded like Audrey would not survive her NICU stay. She was turning blue when the doctor tried to feed her a bottle. She needed oxygen. She had "strange features", such as uneven ears. We'll never forget the doctor that broke the news to us in the worst way possible. We now call him "Dr. Death." In the next two weeks at the NICU, we met many kinder doctors and nurses, as they researched Audrey's condition.

We faced each problem feeling the pain, but still hoping that everything would be okay in the long run. Audrey had some small holes in her heart that the cardiologist told us would probably grow closed. If not, there was always the possibility of surgery down the road, which has a high success rate. She needed oxygen. Okay, she would grow out of that. She wasn't eating very much. But it was just barely enough. When we got the prognosis of Aniridia, we were a little more heart broken. How could our baby



have any *long term* health problems? Then, we read more about Aniridia and decided we could handle this. Lots of kids with Aniridia even go to college! There are eye surgeries that are improving all the time. We would get past this.

Finally, at the end of our stay, the doctors pulled us into the family room to have "the talk." Our little girl has a chromosomal disorder called WAGR Syndrome. As they described the worst case scenarios, we broke down again. All of our building hopes and expectations had been dashed again. This was serious. This was long term. This would not go away. But when we looked at our little girl, held her and loved her, we knew she was unique, different, and not easy to figure out....but she was our "perfect" little girl, just as she was meant to be.



Things might not be normal, but this is OUR new normal and we continue to get used to our new world.

When we took Audrey home, she

needed to be on oxygen. We carried that tank with us everywhere we went. First, the oxygen company gave us huge tanks that weighed about as much as four Audreys! Then, we found out we could get smaller more manageable tanks. While it was never 'pleasant' to have oxygen hooked up to Audrey at all times, it became tolerable.

The next challenge was feeding Audrey. She didn't want to nurse much. The bottle was better, but she still turned it down before she'd had the amount doctors said she needed. Through a swallow study, they x-rayed Audrey's throat as she swallowed and found out she was silently aspirating. We were told Audrey would need to be fed through a g-tube placed in her stomach. Otherwise, she would suffer from liquid in her lungs, resulting in pneumonia. This was a BIG deviation from our "normal", but with time it, too, became part of our routine. Unfortunately, since we weren't giving food to Audrey by mouth anymore, she lost all interest in food and even grew an aversion to it. Now she is not aspirating anymore, but we are faced with the challenge of getting Audrey to *want* food by mouth. We go to weekly feeding therapy and practice eating by mouth four times a day. Audrey will eat a few bites each feeding. It seems she is interested, only because she knows it makes us happy. Feeding is a daily challenge, and the most difficult one we've had so far.

Our experience with Audrey has kept us in a balance between hope and acceptance. While I want to remain hopeful that Audrey will become a great eater, I also don't want to expect this and feel frustrated when it doesn't happen. I want to accept her taking a long time

to learn how to eat, but I also don't want to give up on her making progress in eating. It is a constant balance for me to attain. I find myself leaning one way or the other most of the time.

It was the same way at Audrey's one year Individualized Family Service Plan meeting at her school. I was feeling so great about Audrey's progress in all areas. (Hopeful) When the specialists estimated Audrey's developmental age at six to eight months, I felt my heart sink. I had to find that balance of acceptance and hope again. But without giving up, of course!

We have found great support from the IWSA website and the WINGS newsletters. I love the inspiring words and stories, which help me, pick myself up when I'm feeling down about things. It's good to know parents who have been through the same diagnosis, and share different variations of our experience. I look forward to getting more involved as Audrey gets a little older. We feel so lucky to have Dr. Han and the NIH research team, who is helping us all understand WAGR/11p Deletion Syndrome better.

Audrey has changed our lives completely. She is teaching us daily about love, hope and acceptance along with the importance of community and a connection with other parents.

Written by Tiffanie Strasser, CO

Tax Deductible Donation

The IWSA is a 501(c)(3) charitable organization. We rely on contributions from private and public sponsors. Please consider donating today.

Embrace the "Blessings" in Your Life



Nikki Hoffman, Julie Dell, Tammie Hefty and Kelli Ordakowski at Moms Night Out - WAGR Weekend 2010

I remember the phone call....Julie had the baby. So the big question was "Was it a boy or a girl?" She wasn't saying....I would have to come up to the hospital to find out. Everything is colored blue...."It's a boy!" My mom and dad had two girls, their first two grandchildren were girls....we finally got a boy in the family. The entire family was thrilled.

The next day....I remember the phone call....it was my mom...."you need to come to the hospital, there is something wrong with Hayden". She wouldn't elaborate but just said that I needed to be there. That 10 minute drive to the hospital seemed to take quite a long time. Just having a baby myself, three months before, I couldn't imagine the feelings that my sister was having. When I got to the hospital, things still weren't very clear as to what was wrong; only that the doctors said something just wasn't right. This beautiful baby boy that brought the family so much joy the evening before had issues keeping his eyes open. I don't recall if at the time we knew he had cataracts, I do remember they were concerned that his ear was deformed which could be the sign of several different syndromes. So now what? What is the next step?

The next step was doctor visits, hospital visits, surgeries, research,

tears, laughter, worry, relief, and most importantly, moving on with life as we now know it and dealing with something called WAGR/11p Deletion Syndrome.

There are so many things I could say about Hayden. Hayden is the happiest little boy that you can imagine and he just steals the hearts of everyone that meets him. He is friendly and will say "Hi" to anyone and if they don't say "Hi" back, that's okay, he moves onto the next person. And he is inquisitive; he always wants to know what you are doing.

Hayden has brought perspective to my life in many ways. For example, I went to the eye doctor the other day for a 'normal' checkup. I cringed when they would put drops in my eyes. As I'm sitting there, I start thinking to myself, "Come on Nikki, Hayden does this everyday, several times a day, and you can certainly deal with it for a few minutes!" The drops were meant to dilate my eyes so they could continue with the checkup. After the drops starting taking affect, my eyes were very sensitive to the light (similar to the effects of aniridia for Hayden). So....this is how Hayden sees life EVERYDAY? Wow!

Every time there is a kidney ultrasound or a glaucoma check scheduled, I am constantly looking at my phone during the day to make sure that I didn't miss the text come in from my sister that everything looks good. Those days are somewhat stressful for me; I can't imagine how they are for my sister and brother-in-law. Such relief comes over me when I hear "Clean Beans" or that his pressures were under 20.

I was watching my sister's kids

the other day and all the kids were upstairs playing. After some time, Hayden came down to see what I was doing (remember I said he was inquisitive) and then asked if he could sit on the couch with me. I was huddled under a blanket and asked him, "You want to sit under the blanket with me? His eyes lit up so we cuddled together on the couch. While it was only for five minutes it was the best five minutes of the night!

The people I work with know about Hayden. There are certain ones who are always asking how he's doing and I love telling them the latest stories about him and what is going on with him.

I have a seven year old and she knows that Hayden has some issues, but it's Hayden, and that's how she knows him. She likes being 'mama hen' to him and his younger brother Jaxon. We've been to three WAGR weekends, two of which my daughter and I have volunteered our time. We were one of several other volunteers that kept the kids entertained while the parents had their meetings and discussions with doctors and researchers. My daughter started about a month or two prior to going to the third WAGR weekend asking the question...."when's WAGR Weekend?" That is all she talked about. She was just as excited as everyone else was to go. She loves playing with all the other kids.

During the last WAGR weekend

Forward newsletter submissions, stories, pictures or ideas to:

Annie Prusakiewicz
P.O. Box 392
Allen Park, MI 48101

E-mail: TheMooZoo@aol.com

I was fortunate enough to go along to Mom's Night Out and spend time with the moms. What a wonderful group of moms....they are truly special people. The topic of the night was "the one challenging thing you are facing and the one thing that you are grateful for". I was in the middle of the group and by the time they got to me, I felt so very fortunate that my child is healthy and felt somewhat guilty that I was not dealing with the things that these moms were dealing with. Those things that I thought were challenging in my life didn't seem to be much of anything compared to the things I heard these 'gorilla' moms talk about. Just the simple things in life can be difficult for these moms. I walked away feeling even more in awe at what these moms and dads are dealing with on a DAILY basis. But they keep going, regardless of what road-blocks and obstacles come their way.

So all you aunts, uncles, grandparents, siblings, friends, embrace these kids. Children truly are a blessing and WAGR kids are no different, they just need a little more attention. And just as important....embrace their parents as they are truly exceptional people.

Written by Nikki Hoffman, PA

Cerys' Visit to NIH

It seems so very long ago that we first heard of the planned research into WAGR Syndrome. Way back in 2005 at the WAGR Weekend, we were strengthened by the knowledge already held by Kim, Kelly, Annie and so many others. It was hard to believe that in such a short period of time they'd also enlisted the help of Dr. Fan for his genetic studies at Miami. It was a dream come true to hear of further planned studies at the NIH and we didn't hesitate in enlisting Cerys as a participant.



Blood draws followed, which Cerys took in her stride. Dad was a little less enthusiastic, not being a huge fan of needles, but humbled by the bravery of Cerys and countless others before her, he had blood drawn too along with Mum. Consent form after consent form followed, and we were beginning to become experts in the workings of Fed Ex International Shipping! This is something you guys in the States take for granted, but convincing our local Doctor of the urgency to have these bloods drawn, rushing them by car to the local hospital lab for centrifuge then sending them half way round the world via Customs was a new one on us, and pretty stressful!

We followed the developments of the study over the next few years, but were beginning to wonder whether Cerys would ever be called as an inpatient. You can imagine my surprise when I emailed Dr. Han in the summer of 2010 seeking an estimated date of admission, expecting her to suggest 2012 or 2013, only to have her say "come right over any time after her birthday (September)"! We were elated but nervous and set about planning the trip.

We decided early on that just

Cerys and Dad would travel to NIH. The journey would involve an International flight then a domestic connection and we figured that the distance and cost were too much for the rest of the family to travel. With hindsight this was absolutely the right decision for everyone.

So October arrived, and if I'm honest, I was really nervous about the trip. It would be the first time I'd travelled abroad without the rest of the family and there were a few tears at the airport. Cerys as usual was completely unfazed by the whole process, and enthusiastically waved goodbye to mum as we disappeared up the elevator to the check in desk.

So what followed was the journey of all journeys! (Remember that Eastern Time is five hours ahead of the UK). First off was a flight of around eight hours from Belfast to New York. The increased security meant that we had asked for a slightly longer stop-over at New York and this proved to be wise as the queues were very long for the (very necessary) security checks. We switched to a smaller plane and after another couple of hours touched down at Dulles. We just missed the NIH shuttle and after calling Shannon at NIH I opted for us to take the bus and Metro to NIH, based on the easy to read map and apparently short distance - big mistake! The bus and trains (2) were prompt and very clean, but having never used either in the USA, it really was a baptism of fire trying to buy the correct tickets at the Metro whilst managing our luggage. Cerys really did her best to help. A big thank you to the welcoming staff at the Metro Centre for their patience with me! So we finally arrived at

NIH around 11:00 PM, which taking into account the time difference with the UK, equates to a journey time of 17 hours without sleep.

The staff at NIH Security were very welcoming and called the Shuttle to take us to the Children's Inn. We had a quick tour of the Inn, had a quick shower and went straight to bed.

The next day we went through Admissions and met Shannon and Amanda who ran us through the schedule. Dr. Han was in San Diego for a conference but still called by later that day to say hi. That was a really kind gesture and we appreciated her taking the time to do so.

The next week and a half were an absolute whirlwind of introductions, tests, appointments and early mornings! Each inpatient has an individual schedule, designed to cover the studies and any other issues that the patient wants investigated.

I won't go into the detail of each test, but will summarise my experience of NIH:

- The Inn and the NIH as a whole were amazing. In the UK we would never have experienced facilities of such a high standard.
- Everyone we had contact with, from the Consultants, through to the Nurses, The Coffee Bar staff, the Cleaners, The Inn staff were incredible people.

Nothing whatsoever was too much trouble for any one of them. We were made to feel so welcome it almost felt like we were the only patients there!

- The number of appointments and procedures was incredible, almost overwhelming, and I would recommend that anyone attending should allow a week and a half to complete them.
- Were it not for the calming influence of Shannon, Amanda and the Chaplain, there were times I thought it was going to be too much for us but we got through it.
- The free time at the weekend was an experience to remember. We took in the bus tour of Washington DC and the Smithsonian Museums. The City is beautifully clean and the people were so welcoming, a real credit to the USA.
- We travelled so much on the Metro we were actually advising other tourists on how to use it!
- Cerys loved how the NIH Police covered the Shuttle after hours; we even got treated to the flashing lights and sirens!
- The Inn was like a home from home and we were so



Cerys & Bryan Stewart with Dr. Han and the NIH team

- relieved to get back there in the evenings.
- At the end of our stay we had an informal meeting with Dr. Han where she talked us through the findings and presented us with a preliminary report. This was followed up a few weeks later with a full report.

So we began the return journey but took a cab to Dulles this time, once bitten twice shy! It was a real shock to the system returning back to the UK where the icy weather had kicked in.

I would thoroughly recommend taking part in the study to others. Think not only of the benefit for your child, but all the others who will come in the future. We've been blessed to have the founding members of the WAGR group, were it not for them there would be no Dr. Han and the NIH study!

My favourite highlights of the trip? Cerys having her pink hairstyle to cover the sleep study electrodes and her doing the "ward rounds" complete with clipboard, stethoscope and hospital uniform as provided by the X-Ray Staff!

Written by Bryan Stewart,
Northern Ireland



Shari Krantz &
Dr. Joan Han

Dear Families of the IWSA,

NIH UPDATE

We are very pleased to announce that we are gearing up to begin Phase III of the NIH WAGR/11p Deletion Syndrome Research Study. Through Phase I of our study (which began in 2006), we have learned a great deal from mailed-in records and blood samples. Through Phase II (which began in 2008), we have learned even more by conducting comprehensive studies on-site in our hospital at the National Institutes of Health in Bethesda, Maryland.

We are extremely grateful to all the families who have participated in these studies. We will be continuing Phases I and II because we still have much more to learn.

Meanwhile, based on what we have learned so far, we will soon be adding Phase III. For Phase III, we are planning to conduct a therapeutic intervention trial to study the effects of a medication that may replace some of the functions of brain-derived neurotrophic factor (BDNF) in patients who have deletions involving the gene that produces BDNF. Our hope is that this medication might be beneficial for weight control, neurocognition, and behavior. We were recently awarded a grant to conduct this study and are now in the process of making all the necessary preparations to begin. Our goal is to launch Phase III in late 2011 or early 2012, so please stay tuned for more details.

For any questions regarding Phases I, II, or III of our research study, please email WAGR_study@mail.nih.gov or call (301) 435-7820.

Sincerely,
Joan C. Han, MD
Pediatric Endocrinologist
Assistant Clinical Investigator,
Unit on Metabolism and Neuroendocrinology, NICHD, NIH

Rare Disease Day 2011 at the NIH

by Shari Krantz

In late February of this year in celebration and honor of Rare Disease Day 2011, I went to the National Institutes of Health in Bethesda, Maryland. A fantastic, full day of presentations and discussions took place, including an excellent talk on the WAGR Syndrome/11p Deletion Study presented by Dr. Joan Han, NIH endocrinologist and study investigator. Dr. Han was

joined by Dr. Lazbawan (molecular biologist, State University of New York), who described in detail the genetic testing methodologies used in our studies. Dr. Han's presentation described the study testing protocols, reviewed general results and findings, and discussed study implications. It was fact-filled, fascinating, and emotionally moving--especially for the parent of WAGR/11p Deletion child and study participant. I was full of pride and hope as Dr. Han took the audience through the first two phases of the study and very enthusiastically highlighted key findings as well as areas that she plans and hopes to focus attention

on in the future.

I had the opportunity to stand up at the microphone and share my thoughts and feelings--on behalf of my daughter and as a board member representing the IWSA and all of our families. After thanking Joan and her team for their interest and dedication, I commented to the 100+ attendees that Dr. Han and her team have given our families hope for the future as well as much-needed information and support related to managing the complex healthcare and social needs for our children.

Other interesting and enlightening presentations included discussions of the "Biomedical Translational Information System", the NIH Undiagnosed Diseases Program, and the Genetics Testing Registry.

All in all, it was a wonderful day and I felt very lucky to have had the opportunity to attend the activities on behalf of my daughter and the families of the International WAGR Syndrome Association.

Farewell Shannon, Welcome Amanda

Shannon Fuhr will be leaving her position as WAGR Research Study Coordinator at the NIH in May. Not only was she an important member of the NIH team, she has been a valuable member of our/IWSA team too! We really are sorry to see her go and we wish her well with her new endeavor.

Amanda Huey, BA, will be the new WAGR Research Study Coordinator. We welcome Amanda and look forward to working with her in the future.

**"Never let what you
cannot do stop you from doing
what you CAN do!"**

Stephen Pierce

Fundraising Committee Formed

The IWSA Board is happy to announce the creation of the IWSA Fundraising group. Three individuals will be working closely with the board to help bring awareness to WAGR/11p Deletion Syndrome. They will also help find/share fundraising ideas that families can support as a way to give back to the organization.

Vanessa Richter is aunt to Aydin Duffy who has WAGR/11p Deletion Syndrome. Her sister Elizabeth Duffy is active with IWSA. She has both professional and personal volunteer experience with fundraising. In the beginning of her career she worked for the American Cancer Society and Bloodsource, the Northern California blood bank, as a fundraiser. Vanessa has also been involved in several non-profits including the Junior League, Leukemia and Lymphoma Society and other local organizations. She is excited to work with IWSA and help lend some of my experience with fundraising.

Keri Haertel is mom to 3-1/2 year-old Katie, who has WAGR/11p deletion Syndrome.

Keri has done fundraising pretty much her whole life. Her family was very active at their church when she was young so they used to do things like spaghetti dinners, rock-a-thons, pancake breakfasts, etc. Keri ran her very first fundraiser last year for Aniridia research - it was a children's benefit concert featuring Mr. Steve from Stevesongs (PBS kids). He is truly terrific and happens to live close by! She raised \$14,000! Her goal was \$10,000. She

appeared both in the paper and on a local morning show!

In Keri's mind, every little bit counts, fundraising doesn't have to be a big, time consuming event, it can be small and low stress!

Our last member of the fundraising group is Leslie Volk. She is the mother to three girls, Ashley, 22 - WAGR/11p Deletion; Brittany, 20; and Chelsey, 16. She is married to John, CRNA for the US Navy.

Leslie attended her very first WAGR Weekend in 2010 and says she finally found the support group she had always been looking for.

Her most recent fundraising event was using the *Can you Spare a Dollar?* sheet. She set the sheet out with a basket of candy along with Ashley's picture and the dollars just kept going in. One idea that she has been considering is to throw Ashley a birthday party and ask for donations instead of presents.

The IWSA board welcomes these three talented ladies and looks forward to working with them.

As Keri stated above, fundraising for the IWSA doesn't have to be a huge time consuming event. Thankfully, we are a small organization so we appreciate any and all size donations, large or small.

Fundraising is an important part for any non-profit organization. The IWSA has always strived to provide services and programs free of charge to any WAGR family, family member, professional, or physician.

(Fundraising cont on page 11)

Hockey and Hope II

Take Your First Step

When it comes to fundraising, or sales, or anything financial really, you always want to see an increase from year to year. Last year, our Hockey and Hope event with the Milwaukee Admirals yielded approximately \$700 for the International WAGR Syndrome Association. So, what more could we hope for than an increase?

Unfortunately, it was even more difficult to find attendees for the game than it was last year! People were going to be out of town, which I hadn't planned on. I thought spring break would be the perfect time when I picked a date. I was very disappointed to have to inform Aaron, our Admiral's representative and initiator of this event, that we only had about eight people attending the game, including Jeff, Evie and myself.



Evie Jo shows off the first puck

As I should come to expect, however, WAGR/11p Deletion Syndrome never fails to surprise me. The silent auction donations were incredible! We had items from the Texas Stars, Chicago Blackhawks, and Nashville Predators. The Wisconsin Badgers Hockey Coach signed and donated 4 items as well. The silent auction yielded about \$1200 this year. Amazing!



What was even more exciting, however, was to consider the fact that many of the parents who are part of the IWSA were told years ago that they would never meet another person with WAGR/11p Deletion Syndrome. As we drove home from Milwaukee that night, I teared-up several times with the thought that we had just been on the radio during

intermission and on the big screen there in the Bradley Center having a conversation about WAGR/11p Deletion Syndrome. Wow! The IWSA started with a few moms who said, "We have to find other people out there! We know there are more!" They wanted to reach out to others and say, "Our lives can be filled with joy! Let's share this joy with one another!"



Jeff Hefty mans the silent auction table

And today, our lives ARE filled with joy. When I read a WINGS newsletter I cry tears of joy over the beauty of our kids, their courage, and our courage as parents that we develop along this journey together. Jeff and I may feel exhausted after our Hockey and Hope events; but it's an exhilarating and rewarding exhaustion which means we have helped our families around the world by raising funds that will help keep our newsletter going to print, help keep our website up and running, and help other families get to WAGR Weekend so we can all keep that joy moving around the world and through our lives.

So, while my mind was fixated on getting bigger results and better numbers as we led up to the event; in the end, it was just the "having" the event that made the most impact. Any step I take out there to ask others for support can never "fail" the IWSA. EVERY step we take benefits the IWSA by generating awareness and stirring up enthusiasm. Keep taking those steps out there, and we will find ourselves on a leisurely walk toward the future; and that future looks brighter every day.



Tammie Hefty, raising awareness for the IWSA

WAGR Weekend Financial Assistance

The IWSA offers financial assistance to families interested in attending WAGR Weekend but may not have the resources or financial means to attend. We continually fundraise and budget each year to ensure funds are available. We encourage our WAGR families to apply for assistance. In the past we have provided financial assistance in several forms, including but not limited to: hotel room costs, gas cards, transportation from the airport to the hotel, airfare or train tickets, or combinations of these.

Applying for financial assistance is simple and confidential. Please send your request to Tammie Hefty at tammiehefty@yahoo.com by April 30th and indicate what type of assistance you need. The IWSA will notify all recipients in May so you can make your plans to attend.

WAGR Weekend 2011 Save the Date

The Prusakiewicz Family will be hosting WAGR Weekend 2011 in Southgate, Michigan, July 15-17th.

The Holiday Inn Southgate Banquet & Conference Center is a fine, full-service hotel in the Detroit Downriver Area. Located only 15 minutes from the heart of Downtown Detroit and Detroit Metro Airport it is nestled in a non-congested area, convenient for travel to major shopping, Greenfield Village and Henry Ford Museum.

17201 Northline Road
Southgate, MI 48195
Phone: 734-283-4400
Fax: 734-283-6855
www.HISouthgate.com

Group room rate will be given for all online reservations upon check in

Room Rate is \$87.99
Limited rooms available - first come, first served
Visit www.wagr.org to RSVP today

Holiday Inn Priority Club members will receive breakfast coupons, to enroll visit:
<https://secure.ichotelsgroup.com/priorityclubrewards/en/us/rewards/join/info.html>

Tentative Itinerary

July 15th - 6-8PM Meet & Greet
8:30-11PM Moms Night Out

July 16th - 9:30AM Group Photo
10-11:30AM Parent Information Session/Children's Activities
Noon-1PM Catered Lunch
1:30-4:30PM Offsite Activity (Cosmic Bowling or Swimming)

July 16th (continued)

5-8PM Pizza Party
8:30-11PM Dads Night Out

July 17th -

Informal Gathering of families, if enough interest

All meetings and meals will take place in the Marquis Ballroom located in the front of the hotel near the restaurant.

(Fundraising con't from page 9)

All IWSA Board Members, Officers and Committee members are volunteers. We are an organization with little overhead so most of the money we raise goes into programs, services and potential research opportunities.

The IWSA provides the *WINGS* and Online newsletters free of charge. They are packed full of personal heart warming stories and up to date medical information. Your family may have received support from the *wagr* list serve during difficult times. Families and professionals visit the *wagr.org* website to have questions answered and to seek out valuable information for their child. Your child may have received a special gift when he was hospitalized or your family may have been awarded financial assistance to attend the magical one-of-a-kind event we call "WAGR Weekend". The IWSA needs your help to continue with its mission to improve the quality of life for individuals with WAGR/11p Deletion Syndrome. All we ask is for our families to give back to an organization that is so giving. THANK YOU!



Special thanks to the March of Dimes Southeastern Michigan Chapter, for providing the IWSA a Community Award Grant for 2011. This money will offset some of the printing/ mailing expense of *WINGS* so that we can continue to provide this to our readers free of charge.

The **IWSA** serves families all over the world in countries, including: Australia, Bavaria, Belgium, Brazil, Canada, Croatia, England, France, Germany, Greece, Ireland, Israel, Italy, Kosovo, New Zealand, Peru, Philippines, Poland, Portugal, Puerto Rico, Romania, Saudi Arabia, Scotland, South Africa, Switzerland, Tanzania, Turkey and The United States of America.

Pirates Come through for the IWSA



Friday, January 21st, was Casual Clothes Day for the IWSA in the Riverview Public School District.

Almost \$900 was donated by the awesome Riverview Pirate staff. Way to go Pirates and **THANK YOU** so much for your support!!!

by Annie Prusakiewicz

On behalf of all our families, the IWSA board and officers would like to thank everyone that has made monetary donations to our great cause. We could not carry out our mission without your support. **THANK YOU! THANK YOU! THANK YOU!**

Moving?



Don't miss out on future issues of WINGS. Please send us your new address.

Email our IWSA Secretary, Julie Dell
hcdtank@yahoo.com



International WAGR Syndrome Association

YES Enclosed is my gift

() \$25 () \$50 () \$100 () \$_____

Your Name: _____

Street Address/P.O. Box: _____

City: _____ State/Province: _____

Zip Code: _____ Country: _____

In Memory of: _____

On behalf of: _____

Mail to: **IWSA**
P.O. Box 392
Allen Park, MI 48101

THANK YOU FOR YOUR TAX DEDUCTIBLE DONATION

International WAGR Syndrome Association
PO Box 392
Allen Park, MI 48101



We are now able to accept online donations through PayPal. Please visit www.wagr.org and click on the donate button.

Deliver to: