



WINGS

WAGR Information, News, Gorilla Stories

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

www.wagr.org
Fall/Winter 2010

To all our IWSA families, friends and donors,

On Sunday, October 17th the IWSA Board of Directors held their quarterly board meeting.

The following IWSA Officers were elected: Catherine Luis, President, Tammie Hefty, Vice-President, Julie Dell, Secretary and Jeff Hefty, Treasurer.

Shari Krantz was elected to another two year term on the IWSA Board and Annie Prusakiewicz was appointed IWSA Chairperson.

Kelly Trout and Rhonda Sena will also remain on the IWSA Board. Kelly, who is also the Medical Consultant and liaison will devote most of her time to key medical issues and research. Rhonda Sena, the Educational Consultant will continue to help our families with educational related issues, to access services, therapies and support.

The IWSA Board, Officers and leaders are looking for individuals

who would like to help grow the organization.

We are looking for anyone who would like to help us carry out our vision to improve the quality of life for individuals with WAGR Syndrome.

Have you always wanted to get involved but didn't know how? Do you only have a limited amount of time to donate each week/month? You **do not** have to live in the United States to contribute to the efforts of the IWSA. We are an *international organization*. We support individuals with WAGR Syndrome all over the world. We will work with your schedule. Committees will be forming soon and we need your help.

For more information on how you can get involved and help grow the organization in the upcoming year, contact Annie Prusakiewicz at TheMooZoo@aol.com or Catherine Luis at CatherineLuis@msn.com

Annie Prusakiewicz

IWSA Lending Library

The IWSA has purchased a few books to start a lending library. Descriptions can be found on Amazon.com. Please take advantages of these new resources. If you want to donate books or borrow a book, please contact Rhonda Sena at

CasaSena5@yahoo.com

Book Titles Available

Children with Visual Impairments: A Parent's Guide by M. Cay Holbrook.

A Parent's Guide to Special Education for Children with Visual Impairments by Susan LaVenture.

The Out of Sync Child by Carol Kranowitz.

The Out of Sync Child Has Fun by Carol Kranowitz.

Growing an In Sync Child by Carol Kranowitz.

Special Education Law by Wrightslaw.

From Emotions to Advocacy by Wrightslaw.



On the Inside

Gorilla Stories -	pg 2-5
WAGR Weekend 2010 Highlights	pg 6-7
Across the Pond - by Jenny Langley	pg 8-9
NIH Study Update	pg 9-10
Our NIH Visit - by Tammie Hefty	pg 10-11

The Highlights Of WAGR Weekend

by Robyn Hughes, MD

My name is Robyn Hughes. I am a museum professional, a German Jewish Coffee House Intellectuals expert and a competitive chess player from the Baltimore area in Maryland. I have a Master's Degree in German Jewish History from Baltimore Hebrew University and a Bachelor's Degree in Jewish Studies with minors in Philosophy and in Hebrew from the University of Maryland at College Park. In addition, I have been a visiting graduate student at Harvard University and I have spent a significant amount of time studying at the graduate level abroad. I have a congenital complex malformation Syndrome that has many features in common with WAGR Syndrome and that has required a significant amount of medical intervention.

Every year I look forward to attending WAGR Weekend because it is the only place that I have found where I can socialize with a group of people who are able to imagine what it is like to be a successful professional and to have to undergo the specific medical procedures that I have had to undergo. Every summer, I eagerly await the opportunities to see long

time friends and to form new friendships. I always tell my Mother that I feel as though I am going to summer camp. A camp that I know well and that I have been returning to year after year for nearly a decade. WAGR Weekend has afforded me the opportunity to network with medical researchers.

In addition to networking opportunities, I also look forward to the lighter moments during the WAGR Weekends. This past WAGR Weekend I immensely enjoyed playing chess and also had a wonderful time socializing at the pool where we explored a giant fountain. This was a particularly welcomed attraction by all of us who had spent time earlier that afternoon walking around the park grounds, as it was over 100 degrees.

Every year I return home from WAGR Weekend happy that I was able to attend the event and always looking forward to returning the next year!

Our First WAGR Weekend

My husband John and I with our daughter Ashley attended our first WAGR weekend in July of 2010. During a deployment, John had found a web site about 11p- deletion, which our daughter Ashley was diagnosed with in 1988. He forwarded this information on to me and I quickly looked into it. At the time of Ashley's diagnosis the doctor said that her chromosome deletion was rare with about 1 in 100,000 live births. She gave us some information and sent us on our way with numerous follow up appointments and referrals. As first time parents of a precious four month old, this type of news is devastating. There were no

groups to support families; there were no support services for this diagnosis. I was very glad we had finally found a support group and that we were no longer alone. Before attending this weekend, I had not met another child with the same diagnosis as Ashley.

What a great time we had at our first WAGR Weekend event. I had found a few people within this group on the internet and talked and asked questions. I had to encourage my husband to come with us for the weekend. I'll never forget how welcomed I felt walking into the lobby of the hotel, the culmination of the many months on the internet. We met so many wonderful people with shared experiences. I felt an instant bond. We were overwhelmed with the generosity of this instant family.

Knowing that it might be overwhelming for Ashley, we kept the weekend at her pace. There were a lot of activities to take in. It took Ashley a bit to feel comfortable with everyone and when she finally did, it was time to say our good-byes and plan for the next year. I wish that we were able to bring our other girls; I think that Ashley would have been more comfortable from the start. The events that were available were great. We only took in what Ashley was willing to do, as not to upset the situation. But as we found out, other families were dealing with the same situations or behaviors that we were.

We took in all the information about the research study, signed up for it and are looking forward to learning more about our daughter. I'm sure that all the knowledge we learn will make our children the best they can be.



Caroline, Jenny, Robyn, Norma & Evie

We are already making plans for WAGR Weekend 2011 and this time we are not leaving anyone home. I can't wait to see everyone again and hear the study updates.

I asked my husband to share his thoughts about the weekend. Here is what he has to say....he isn't usually too much for words.

Warmest regards,
Leslie Volk, CA

Eye Opening and Rewarding

I was originally going to let Leslie and Ashley attend WAGR Weekend and then determine through their feedback whether or not I would attend in the future, a mistake I am glad I did not make. This weekend was one of the most eye-opening and rewarding experiences of recent memory.

Ashley is twenty-one years old and we have navigated the trials and tribulations of dealing with a developmentally challenged daughter without the knowledge of her specific challenges or needs. We knew that we would have some difficulties after speaking with the dysmorphologist while Ashley was an infant but the challenges grew



Ashley, John & Leslie Volk

and evolved in their complexity over the years. With time we developed strategies to minimize the difficult situations but still struggled with many behaviors.

It was inspiring to spend time with a group of families and their children, challenged and not, and to see that similar challenges are experienced by all. Even more inspiring are the siblings of our special children and the love and understanding that they exhibit. My only regret is that our two younger daughters were not able to attend.

A most memorable experience that I will definitely look forward to again. Thank you for sharing your families and your time.

John Volk, CA

Gorilla Grandparents

We started coming to WAGR weekend in 2004 after our grandson Hayden Dell was diagnosed with WAGR. We were both anxious, nervous and excited to come to the first meeting because we didn't know what to expect. But once we met Catherine, Kelly and Annie we were put at ease and became one big family. We are learning a lot and are coming to understand it better.

We are so proud of Hayden's parents, Julie and Brian. Not once during the time of Hayden's diagnosis, operations, therapies, sonograms, EUAs, doctor appointments, setbacks and accomplishments have we heard them say "why us?"

God saw something special in them and that is why he gave them Hayden.

So now six years later we still

Steve & Monica Wright



enjoy coming to see our second family, old and new alike. We are always amazed how these kids have developed and have come so far.

We are also happy to see what a big support group these families have become to one another whether it by phone, email or Facebook. WAGR Weekend has become a highlight to our summer.

Steve & Monica Wright, PA

New WAGR Grandfather

I recently became a grandfather for the first time to a five year-old daredevil named Aydin Duffy. Aydin has WAGR Syndrome which I had never heard of until about a year ago. I didn't know Aydin when she went through her eye and kidney surgeries and chemotherapy during her first few years of life. I can only vaguely imagine, thank God, the pain and worry her mother Elizabeth must have gone through.

Forward newsletter submissions, stories, pictures or ideas to:

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

E-mail: TheMooZoo@aol.com

When I first met Aydin-with those coke-bottle glasses of hers-I marveled at how she maneuvered around our house and backyard, at full speed, with eyesight equivalent to legal blindness. Her imagination was never-ending and her imagination that took me back to my own childhood. She challenged me and my imagination into wondering about all the possibilities a day could hold with just a few sticks and leaves to play with and some grass to run on. In a matter of an afternoon we would both become pirates, sailors, whales, dolphins, kings and queens, princes and princesses, dragons, moms and dads, brothers and sisters....you name it and it just seemed to happen.

When my wife and I were invited to our first WAGR Weekend by our son Nathan and his wife, Elizabeth this last July, we really had no idea what to expect. Within two hours on that first Saturday morning, Brenda and I learned more about Aydin's special world than either of us had been able to put together on our own in more than a years' time. I began to understand some of the real physiological and psychological challenges that come with living with WAGR. A wonderful physician named Dr. Han, who gave up her whole weekend to be with our families, described many of the WAGR

projects being worked on at the National Institute of Health. Another physician described current advances in eye surgeries. I heard from several parents of WAGR children and saw how the families encouraged the kids to be a part of the weekend. I also gained a greater understanding of how this small group of families had organized themselves and how this weekend event was so very important for their encouragement each year.

I got to spend an evening with a few of the fathers and grandfathers of these special children and hear about the personal difficulties and blessings of raising WAGR kids. I learned about the important life-transition stages of WAGR kids from these dads. I also learned about the legal ramifications concerning special-needs children who become adults in the eyes of the court.

But the most impressive and important thing of the weekend was meeting a dozen or so of these children, at differing ages with differing aspects of WAGR symptoms. The gym was the best place to do that as the kids played all kinds of organized and unorganized games. This allowed someone like me to interact with the kids and better observe and understand some of the similarities and differences of WAGR children....and it also

allowed me time to see some of their rather wonderful traits as well.

These kids are scrapers and tougher than most. They are both brave and fun. They seem to

love people intuitively and seem more naturally inquisitive than most. Most are not shy and seem to overcome their handicaps by other means. They may not be able to see well, but they can hear very well. They may not be the most athletic kids but they make up for it with joy, effort and determination. They love the game of life, from top to bottom, even though they have had sizeable barriers put in front of them from the start. But don't tell them that, because they don't see it and won't know what you're talking about.

Mike Duffy, CA

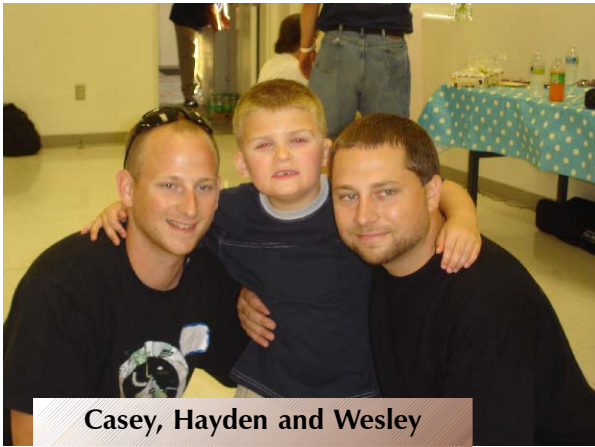
Part of the WAGR Community

WAGR Weekend 2010, held in Gaithersburg, Maryland, was the second IWSA event my family has attended since my sister, Amy, and mother Shari Krantz became involved many years ago. My wife Brittany, brother Casey and his girlfriend Charity, and I had the privilege to host the gym activity time with all the attending children. After the second year in a row we entertained the kids in the gym, we once again came away astonished at the capacity our WAGR children have to deal with adversity. If working with children keeps you young, working with WAGR children gives you the ultimate perspective on life. Before our first visit last year I was not sure what to expect, and quite honestly was nervous about hosting the gym time. As soon as the children began to come in, the reality of the situation set in. One by one my worries and preconceived notions of what it was going to be like were out the window. With the smiles, jokes, please and thank yous, and the bas-



Aydin, Hayden, Mike & Jalen

Siblings - A Special Connection



Casey, Hayden and Wesley

ketballs flying all over gym there was no time for me to feel bad or awkward because the kids were too busy having fun. The joy that came from a couple hours in the gym is amazing and the relationships with the kids that a year later not only remember your name but exactly what you did last year, give you an understanding as to how important this weekend is for the families. We were thrilled to play with all the kids again, and especially enjoyed seeing how many of them had grown and matured.

When my wife and I talked on the way home about the day, we agreed that we both felt privileged to be a part of the WAGR community. This past year served to reinforce for me the importance of not only WAGR Weekend but the ISWA organization in general. In the same way we all can find comfort in our family and friends, my family has found comfort in being able to interact and spend time with other families who can understand our unique struggles. WAGR Weekend offers that opportunity not only for the parents but also gives our children a rare opportunity to be amongst peers who experience some of the same things they deal with on a daily basis.

We are looking forward to next year and continuing our new friendships.

Wesley Marshall
(brother to Amy Marshall, WAGR, 15 years old, Maryland)

I enjoy attending WAGR weekend every time that my family is able to go. Everyone connects so easily there, even if you don't know somebody you are almost automatically friends with them because you can relate to them so well. I think WAGR Weekend is great because you can find at least one person that you can just talk to about anything, without knowing them for 24 hours. When I see the other WAGR kids I feel very lucky, because my sister, Kelsie is extremely happy and way easier to put up with than some of them. I feel guilty though, because I see

WAGR kids that are way worse off than Kelsie, and their families are great with them, but I fight with Kelsie. My favorite part was all of the fun things that Ashley and I did together, I can't pick just one part.

Rockie Brooks, IA
(sister to Kelsie, 18 years old)

Every year I look forward to WAGR weekend. It is a place where everyone is the same and you don't get looked at funny when your brother is screaming or going through another meltdown.

When you are at WAGR Weekend you don't have to feel alone, because you are not the only family that is going through

the meltdowns, temper and anger problems. WAGR weekend is a time when you can connect with other siblings that share and endure the same things as you.

This year I was very lucky to spend time with Rockie. I loved that we could have fun together and share stories about our siblings. WAGR Weekend is a great way for families to connect with one another and to feel at home. I am very excited that my family is hosting WAGR Weekend again in 2011 and I hope to see everyone there!

Ashley Prusakiewicz, MI
(sister to Nicholas, 14 years old)



Rockie, Dalton & Ashley

Tax Deductible Donation

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

Memories from WAGR Weekend 2010

Dads Night Out— (back) Brian, Clem, Mike, Nathan, Tom (front) Gerald, Jeff, John, Steve



Jenny & Evie



IWSA Board Members, Officers & Leaders



Moms Night Out (back) Kelly, Norma, Elizabeth, Brenda, Catherine, Shari, Connie, Leslie, Kelli, Rhonda (front) Robyn, Kelli, Tammie, Julie, Annie, Nikki



Celebrating Hayden's Birthday



Aydin & Maddie



Amy, Jenny & Irma



Kelsie



(back) Sebastian, Darius, Alex, Dalton, Irma, Jenny, Caroline, Robyn (front) Amy, Evie, Kyle, Hayden, Jenna, Nicholas & Kelsie



Jenna, Nick & Hayden's annual elevator ride—Going up?



Kelsie with Dr. Han and Emanuel



The Ordakowski Family



Charity & Hayden

WAGR Weekend 2010 Sponsors



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- Tim & Deb Travers
- Dave Vidmar, State Farm Agent

WAGR WEEKEND 2010 DVD Available

Important and interesting information was presented at WAGR Weekend 2010 by Dr. Joan Han and members of the WAGR Syndrome/11p deletion study team. Dr. Han provided an overview of the WAGR-related studies currently underway at the National Institutes of Health as well as specific find-

ings and trends. Dr. Zein, one of the NIH ophthalmologists involved with the WAGR studies, discussed aniridia and glaucoma-related issues in individuals with WAGR Syndrome. Our IWSA medical consultant and IWSA board chairperson, Kelly Trout, presented an informative and moving video presentation illustrating the history our of wonderful IWSA organization and its families.

If you would like a copy of the WAGR Weekend 2010 DVD, please contact Annie Prusakiewicz at TheMooZoo@aol.com. This DVD is free upon request. Donations to offset the production and mailing expense are appreciated.

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

Room Rate is \$87.99



ACROSS THE POND TO WAGR WEEKEND

by Jenny Langley

On Thursday the 22nd July, I set off on my second journey across 'the pond', this time to attend WAGR Weekend 2010 in Maryland.

The flight went well, although it felt like it took forever! Once off the plane, I was met by a woman with a rather abrupt attitude who shoved me into a wheelchair and didn't listen to my protests! I went on the funny bus that looks like a living room inside and was then taken to customs, where I had my fingerprint taken and my (non-existent) irises scanned. They must have fast tracked me because I didn't wait in a queue, which was great. After my bag search it was to the exit. The driver (Amy's uncle), was lovely and gave me a bottle of water. I was instantly hit by the heat when I stepped out of the airport, it was amazing! We arrived at the hotel, I checked in and was taken straight to my room. I was absolutely amazed--the room was MASSIVE--it had a kitchen with a microwave, coffee maker and fridge, and a living room area.

I had just settled in when the phone rang. It was Shari Krantz (Maryland) saying they were down in the lobby with some food for me. Shari's daughter Amy came up to collect me because I wasn't sure which way to turn out of my room to get to the elevator. I was so happy to see Amy again and get a big hug from her. I went downstairs and met up with Shari and three other families. I sat with Rhonda Sena (New Mexico) and her son Alex and opposite Irma Luis (New Jersey) and Amy. Alex kept telling me when I'd missed a bit of dinner, which made me laugh because usually I'm self conscious about eating in front of people but Alex's help put me at

ease--thanks Alex!

Nicholas Prusakiewicz (Michigan) also put me at ease by shaking my hand and being ever so polite when he met me, what a gentleman he is! When I finished my meal, Amy kindly took me back to my room and showed me around the kitchen and the bathroom. I went to bed very early, as I was exhausted from travelling.

On Friday morning, I got up, relaxed and tried to recover from jet lag! I wandered down to the foyer at 1pm and this was perfect timing because several families were there and more were arriving. I met the Dell and the Cox families (both from Pennsylvania). Next I met Kelly Trout and her daughter Caroline (from Texas)--wow, what a moment! It was surreal to meet Kelly after 'talking' to her online for nearly ten years. From the moment I met Caroline, she (along with Irma) looked after me so well. Later on Friday when all the families had arrived, we had the 'meet and greet', which I enjoyed. I had some more yummy food, visited more with Caroline and Irma, and met some other families. I also met Tammie Hefty and her daughter Evie (Wisconsin)--it was great to meet them after talking to Tammie online for several years.

I was up early on Saturday morning and got a lift with Kelly to the park for the day's activities. I sat with Caroline and listened to the presentations from the IWSA and Dr. Han and her team from NIH. It was interesting and good to hear what the research study is finding. After a nice buffet lunch, I got changed and headed out to the water park. Everyone else was complain-

ing about the heat, but I thought it was amazing and loved it! I made the most of it, as we don't get those sorts of temperatures over here in the 'chilly' UK.

I played under a big mushroom waterfall with Robyn Hughes (Maryland) and enjoyed meeting Jenna Cox. I also got a quick cuddle with Evie. Wow, that was a dream come true! I was so happy I finally got to hold her in my arms, after following her progress for so long. I was truly touched when Kelly asked Caroline and Irma what their highlight of WAGR Weekend had been and both girls said "Meeting Jenny"! Saturday evening's pizza party was AMAZING and we all had a chance to visit some more.

On Sunday I enjoyed chatting to Robyn and got a proper cup of tea too, with thanks to Robyn's mum Norma. I enjoyed chatting briefly to Elizabeth and Nathan Duffy and their cute daughter Aydin (from California).

All too soon, it was time to leave. Amy's uncle took me to the airport and ensured I was with a member of staff before he left. The plane was delayed because of a big storm that hit Washington, DC that evening, but thankfully it didn't affect my flight too much. Once I got to the bus station back home I got a lovely welcoming party - my mum and Jaynie!

Thank you to everyone who made it such a fantastic weekend for me! Thank you Shari for ensuring that from the minute I arrived in the US to the minute I left it, I was well looked after; thank you to Amy for all the help you gave me; thank you to Caroline and Irma for being so friendly and instantly accepting me as 'your friend'. You are two AWESOME women and I love

you-hope to 'chat' to you again soon. Thank you to Kelly and Caroline for my new Texas long horn cap! I love it and will always think you both when wearing it. Thank you to all the families who helped me, everyone was so lovely and nothing was too much trouble for anyone.

There were so many memorable highlights from WAGR Weekend 2010, here are a few for me:

Rhonda saying "Oh it never occurred to me that you would have a British accent ... that accent never comes across on the email!" So funny!

The total feeling of inclusion and acceptance, and no one asking "How much can you see?"

The surreal feeling that everyone seemed so familiar even though I hadn't ever met them. I felt instantly comfortable with everyone as if I'd met them hundreds of times before.

One family saying "Oh you're Jenny?! We read your emails on the listserv and like following your progress." A nice reminder that even though not everyone personally replies to emails, many are reading them and praying for or thinking about you.

Those sudden moments of total realisation-- I like wearing my swimming costume, I wonder if that has to do with sensory

processing?' I asked Tammie "Does Evie like wearing her bathing suit?" and she replied "Yes, I guess it's because it's tight fitting." - amazing!

And, my only regret? Not having enough time--there were some families I didn't even say 'hi' to, which was a shame, and some families that I wished I was able to spend more time with.

I would love to come to another WAGR Weekend in the future but my trip also spurred me onto thinking about planning a similar event for WAGR families in the United Kingdom.

NIH UPDATE

Dear WAGR Families,

We are happy to report good progress with the WAGR Syndrome Research Study at the National Institutes of Health (NIH) in Bethesda, Maryland.

First, we would like to introduce

ourselves, both existing and new members of the research group. Joan Han, MD is a board certified pediatric endocrinologist and the principle investigator for the WAGR study. After attending college and medical school at Harvard University, she trained in pediatrics at Boston Children's Hospital and in pediatric endocrinology at Nemours Children's Clinic in Jacksonville, Florida and at the NIH. Working along side Dr. Han is Shannon Fuhr, BA, the research assistant coordinating the WAGR study. She graduated from Vassar College in New York with a degree in Neuroscience and Behavior with a focus on pre-medical studies. To assist Shannon, we recently added another research assistant, Amanda Huey, who just graduated from Williams College in Massachusetts with a joint-degree in Chemistry and Economics, with a focus on pre-medical studies. For those of you who have met Mark Lee, our former student volunteer, he has begun college at University of Maryland this fall. In his place, we have two new student volunteers, Jamila Crossman and Matt Tsang, both from Walt Whitman High School in Bethesda, Maryland.

We are continuing the Phase I stage of the study and are making significant advancements in Phase II, which would not be possible without the remarkable and much appreciated involvement from the wonderful families of the IWSA. We have had 57 families from all around the world participate in Phase I, which launched in June 2006. In Phase I, participants of any age are involved in a mail-in study. This includes a blood draw performed at your location and collection of medical records. One or both parents may also choose to have blood drawn, but this is completely optional.

We have already had 26 families participate in Phase II, which launched in November 2008. Phase II is an option to come to

WAGR Syndrome/11p Deletion is a very rare disorder. Physicians are generally not familiar with this condition, or with the features and complications of it. People with WAGR Syndrome benefit greatly when family caregivers, medical professionals, teachers and therapists learn as much as possible about the disorder, and become active partners in their care. "WAGR" is an acronym. The letters stand for the most common features of this disorder.

W-Wilms' tumor

A-Aniridia

G-Genital and/or urinary tract abnormalities

R-Cognitive delays/developmental disabilities

People with WAGR Syndrome have many things in common, but they are also individuals. It is important to remember that a given individual with WAGR Syndrome may or may not have or develop the same conditions.

the NIH for evaluation. For participants age six years and older, the study consists of a week-long inpatient stay, during which a comprehensive evaluation is conducted to explore the relationship between gene deletions (genotype) and clinical symptoms (phenotype). The studies include blood and urine samples, radiology imaging, and examination by multiple doctors specializing in various fields, including nephrology, ophthalmology, endocrinology, neurology, psychology, audiology, dental, and psychiatry. After your stay at the NIH, all of the results are explained and sent to you as well as to your child's doctor.

For children age two to six years old, there is now the option of participating in an abbreviated version of Phase II. This visit would involve several days of outpatient evaluations at the NIH, which will include a fasting blood draw and neurocognitive testing. Limited non-research tests may also be performed based on clinical necessity. You will receive the results of these tests and can choose to come back to the NIH again after your child is six years old to complete the rest of Phase II.

What have we learned so far from our studies? The following findings have already been reported at scientific meetings and in medical publications:

- 1) Deletion of the brain-derived neurotrophic factor gene, *BDNF*, increases risk for developing childhood obesity.
- 2) Deletion of *BDNF* is associated with higher pain tolerance.
- 3) Larger deletion size is associated with shorter adult stature.
- 4) Patients with WAGR Syndrome share some common atypical facial features.
- 5) Pancreatitis can occur in WAGR Syndrome, particularly if triglycerides are elevated.
- 6) Using only an interview-based method to diagnose autism in patients with WAGR Syndrome may not be accurate because many patients

who screen positive on the interview, turn out not to have autism when using a full observational assessment.

We have also had many other important preliminary observations that we are exploring further, including auditory processing abnormalities, defects in eye movements, behavioral and psychiatric problems. We are very appreciative of the information that the participating families have provided and invite more families to join our research study.

If you would like to participate in Phase I and/or Phase II of the study or if you have any other questions about research at the NIH, please feel free to contact Shannon or Dr. Han.

Shannon Fuhr, BA
(301) 402-6762
WAGR_study@mail.nih.gov

Joan Han, MD
(301) 435-7820
hanjo@mail.nih.gov

A Mission, Not a Vacation

During our week at the NIH for Evie's participation in Phase II of the study I realized that I had built up some preconceptions of what would be happening. Those preconceptions were big obstacles for me as I searched for peace throughout the week. I wanted to share with other families who may be planning to participate so they may have an easier time adjusting their preconceptions to reality.

My Preconception: Because we were taking a trip, and I was taking vacation time to do it...we were on VACATION.

Reality: There is nothing about the study that qualifies as a

vacation. You are on the go, you are mentally taxed at times, and your child is truly under a microscope. About halfway through the week, I realized that Evie was still equating the trip with vacation, so I told her that we should really call it a MISSION. I told her that we were there to make sure her doctors and teachers had the best ideas about how to give her medicine and how to teach her, and the people at the NIH could tell us that.

My Preconception: We would be staying at the Children's Inn, so we wouldn't feel so "hospitality". I pictured the appointments really be more like "clinic" appointments, not realizing that I would be admitting my daughter into a hospital.

Reality: Most of the time (7am-5pm) during the day is spent in the hospital with an assigned hospital room as a touch-base-meeting room of sorts. The participant has to be admitted to the hospital in order to get the meal plan. Also, there are at least two over-night stays depending on the sleep study and arrangements of other tests. So, prepare your child that there will be lots of time spent in the hospital.



My Preconception: Everyone at the NIH will know that we are there for research, and everyone will know what WAGR Syndrome is. I don't know why this is what I thought, but I really did think that we were going to walk into a world where the people would be grateful to us for participating in research and that they would all know that we were there, not because our kid was sick, but because we were trying to help everyone know more about WAGR Syndrome.

Reality: I'd say about 50% of the people who we came in contact with knew that we were there because we were participating in the 11p Deletion research project. Most of the WAGR Syndrome technicians didn't know anything about why we were there, so we were kind of treated like we didn't know much about why we were there either.

This frustrated me because I had created such an "expert" status of all people associated with the NIH, that it stunned me when EVERYBODY didn't know EVERYTHING. I have gotten so burned out on bringing new healthcare professionals here in Madison up to date on our history and what WAGR Syndrome is, that I didn't want to be



hashing through all of that again while I was at the NIH. But, in some cases, I did have to go through it all again.

My Preconception: Because this is the NIH, we won't have the same kind of mis-haps that we have at the hospitals and clinics in Madison. I won't have to be "on my game" as much as I am back at home.

Reality: I had to be "on my game" because I still was the expert on my child. For example: There was a point in the week where Evie was going to be fasting four mornings in a row. I looked at the schedule and didn't really think anything of it. However, when we got into that stretch of fasting, I suddenly realized that she wasn't getting her morning meds, and then the day was so full, that we were not getting her the meds soon enough to space them out before her nighttime meds.

I finally had to call off the fasting after two of the four days because it had gotten to a point where I was no longer comfortable with the thrown off medication schedule. Everyone was very understanding, but it was still frustrating to me that I had fallen short on foreseeing that problem when I had been in possession of our tentative schedule for weeks.

This summarizes a few of my biggest mental challenges while we were at the NIH participating in Phase II of the study. There are several other areas that I found challenging, and I plan to write about all of them over time. I am so glad we participated and we came home with a number of recommendations for ways to improve Evie's quality of life from new goals for blood pressures, to new ideas for how to strengthen her hips and leg joints to make her a more stable walker. I recommend that everyone consider the possibilities that can come out of participation in the WAGR Syndrome/11p Deletion study at the NIH.

Tammie Hefty

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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.

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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.




You Got Lemons....

If you got lemons....why not make some lemonade?

*Alanna, Caitlyn, Madalyn and Owen wanted to do something for a good cause, so they decided to have a lemonade stand on a hot summer day and donate the money to the IWSA in honor of their friend Hayden. The kids set up their stand in front of their house and glass by glass they profitted \$20.50. **Thanks kids!** You may be little, but you sure have big hearts!*


Julie Dell (Hayden's mom)

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!

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