



WINGS

WAGR Information, News, Gorilla Stories

International WAGR Syndrome Association
P.O. Box 1346 Manassas, VA 20108

www.wagr.org
Fall/Winter 2007

The President's Corner

Dear Families & Friends,

Needless to say the anticipation of WAGR Weekend each year never loses it momentum for us. This year was a great gathering of new families & old friends. Many thanks to Dr. Joan Han and Ana Morales, MS, for volunteering their time to present updates on the WAGR syndrome research projects.

After meeting all you it left me asking what can IWSA do to improve our method of empowering each family and health care provider?

We've revamped our website with focus on education about the different diagnoses that sometimes occur in addition to the W-A-G-R. Our new WAGR Lite page reflects how beautiful and wonderfully funny our children are. We go through some very tough times and when our children smile or clown around it melts away the stress.

A new Welcome Packet has been created on CD to simplify access to information. This enables new parents to digest it at their own pace & share with physicians as needed. Also available on request is a Physician Information Packet that can be sent directly to any physician involved in your child's care. Sometimes parents are uncomfortable doing this so we will gladly do it

for you. Our brochures have been redesigned and updated. We're enclosing a few for you to share. Please contact us for additional copies.

WAGR Research Presentations are available via e-mail or on disc for families upon request.

No one likes to talk about the "M" word, yet money is necessary to keep our organization running and our materials free, as well as to fund WAGR Weekend meeting space, meals and travel assistance to families. We do apply for grant money to help us with some of these expenses, but we never know what funding we will actually receive.

- Our latest fundraiser for IWSA is our CaféPress store. You can access it from our website or <http://www.cafepress.com/wagr>

They do ship internationally.

- Did you know if you shop online you can automatically donate to IWSA? Just sign up and you're on your way! <http://www.igive.com/welcome>.

- Please use GoodSearch as your main search engine from now on and tell your friends, family and co-



CaféPress Merchandise & Apparel

workers so they, too, can start using GoodSearch to support our organization! Please visit our donation page www.wagr.org and review all the different ways you can help.

Catherine Luis, IWSA President



Jenna Cox & Nicholas Prusakiewicz

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Gorilla Stories

Tears come to my eyes as I start to think about writing this. My undying devotion and love for Wesley makes it hard to write about the bad times. Because all I know is that we have a wonderful, loving grandson that has brought more joy to our lives than words can tell.

Wesley Adam Schmer was born in July of 1995 to Adam and Olivia. He has two younger sisters, Madison and Isabella. Twelve years ago Adam and Olivia took home a very healthy and hairy little guy. At about 6 weeks of age and after what we now call night terrors, (many of our children in WAGR have been through the long endless crying at night, not to be confused with colic) both grandmas insisted a very unconcerned doctor tell us why his eyes didn't seem to look right. Egotistically he said we were just an overprotective mom and grandmas. Well, sure enough he found Aniridia and immediately called a very good pediatric eye doctor. Within two months Wes was in surgery. His cataracts were already so bad that they decided they had to remove them then. He has gone through many, many EUA's (Exam Under Anesthesia). We were relatively free of eye drops on a regular basis until a few years ago and now Wesley uses them morning and night. He has had what I call a little tube in both eyes surgically inserted to help keep the glaucoma under control. They have done a great job for him. Wes wears thick glasses but has very good functional vision. He is starting to learn to use a cane at school, is working on learning to use a Braille writer, and has a magnifier to help him read.

For reasons we don't know yet, Wesley has not had Wilms' but we still have him checked every year anyway. We found out at 2 months that doctors don't know everything! His ultrasound as of August 29th, 2007 was normal!

Wesley has had one testicle re-

moved as it looked suspect and the other is not developed so they continue to watch it. As far as the R, yes Wesley is developmentally delayed. He has some autistic characteristics in that he repeats things and sometimes fixates (by talking and talking about them) on events or things. Otherwise he is most unautistic in that he talks a mile a minute and is the biggest social butterfly.



Wesley and Madison Schmer

At his school we can't even go down the hall without nearly every child and teacher saying, "Hi Wes". He has always been in special education at a regular education school. This past year he would go out and into a regular 6th grade class where he would do his math (about 2nd grade) while in there. He also went out for science where the teacher loved having him because while the others wouldn't ask any questions he had a million and one. This only happened because of kind, patient teachers that wanted to be a part of helping Wesley grow academically and in peer settings. He was only allowed to go if his behavior was appropriate. I think he only had to give it up a couple of times. This year he will go into a self contained special education class in junior high with lots of peer teaching. Again it is a school that is very use to and accepting of special needs children. Our district is suing the state

right now because of lack of funding for special needs students, so everything is changing in special ed. in our district. To the extent that many services could be done away with!

Now I have covered the WAGR part of Wes let me tell you little something about this young man. Some of you got to meet us when we came to the second WAGR weekend, or the aniridia conference in Tennessee and the WAGR weekend IV in Tennessee. We live in Washington State. I am the proud grandma to Wes but I have him living with us at least 3 or 4 days a week. He gets a lot of the special work he needs on everyday things and lots of exercise as we have a paved trail that goes for miles. Yes, like many he has constant weight issues. The exercise works for grandma cause I do too and he will get me going on days I would rather not and I get him going on days he would rather not. When we do our walks we will usually go for about 3-5 miles. Wes can truck with the best of them. He always amazes us. I am sure if you have a little one, you like us, wanted to know how far can my baby go and what will his future be? All I can say is that we assume he will do it all and when there is something he can't I look to God and say "Thank you, for all you have let him do".

Because everything is delayed, I remember how we wondered if he would talk. Now we can't keep him quiet. Would he ever read? Wes loves to read, (only at about 2nd grade level but how thankful we are). Would he ever eat without getting food all over him? Well, that is still a work in progress. Would he be able to ride a bike? We started him on the beach at about seven and now has graduated last year to an adult two wheel bike (rides with his papa at least once a week on the trail to their favorite places). Would he be able to play sports with such poor vision? He has played special ed baseball for about five years and has skied for the past 6 years with skiforall (it is volunteers that work one on one with developmentally

and physically challenged children and adults). What a joy it is to watch him ski down, (slowly mind you) without assistance but always with an adult right near.



Wes is an outside boy. He helps to clean out his goat pen, keeps the water filled and takes turns with his papa feeding his goat. Now mind you cleaning out the goat pen is shoveling all the shavings into a wheelbarrow (I dump so it doesn't get dumped all over the ground) and sweeping it out. I let him wash the car, which he can do for hours. I figure by the time he has washed it three times its pretty clean. Years ago he took a real interest in taking pictures so I thought ok we will give it a try so I bought him a disposable camera. For every 12 pictures one might have something in it. We decided that was not for him. When I got my digital camera, I let him use it. He got so good that for his birthday his Aunt Becky, and Uncle Matt and family bought him his very own. He has gotten some priceless pictures. His aunt insisted on getting the extended warranty, which was wonderful because we took it hiking and he fell face first with it around his neck. However, we got some great pictures from the top of the mountain.

Wes is very into wanting to be independent and we work on that but outside of home it is still really hard. He has a long way to go on appropriate behavior. He thinks everyone is someone to talk to and is still not aware enough of his total surroundings. We try things like letting him walk on the trail where he wears a watch and must be

back by ten minutes or he can't go again for awhile. I broke down and followed him to be sure he was ok. I can just move by trees and he doesn't catch me watching. Guess that's a sight for trail goers and I am glad no one has reported me for stalking.

I hope that by telling my story, you can not only see who Wes is but also how far he has come and how much we have to look forward to. Yes, we will have surgeries and constant eye appointments and maybe even some unthinkable hurdles but we are not alone. Others have gone before us and have been able to get through it. I don't think for one moment that there might come a day when something will happen that will take him away, but that can happen with any of our children. I can only continue to give him all the tools he will need to live as independently as his life will dictate. As his grandma, my hope is that when I leave this world I will have peace in my heart that Wes's life will be good.

God bless each and every one of you and your families. I don't post often, but I read and you are all in my heart and in my thoughts. If ever I can help by sharing or listening or if you have questions about Wes feel free to email me at bevschmer@hotmail.com. It would be my pleasure to hear from you.

Written by: Bev Schmer

When my son Alex was born in May of 1992, we had never heard of WAGR Syndrome. He'd been given the diagnosis when he was 3 months old, and we spent the years following slowly learning what that meant. Many of his

**Health Questions?
Families & Physicians please
Contact:
Kelly Trout, RN, BSN
IWSA Health Consultant &
Liaison to our Medical
Advisory Board.
KellyTrout@sbcglobal.net**

physicians knew nothing about the syndrome at the time, and www.WAGR.org would not come into existence for another 6 years. It would be 5 years before we would have any contact with another WAGR family, and 2 more after that before the first WAGR Weekend, which was attended by a grand total of 7 families. I am relaying all of this so that you will understand why what happened in October of 2000, when Alex was 8 years old, was so mysterious to us at the time.

One Thursday night in October of 2000, Alex started to complain about abdominal pain, and then started vomiting. We assumed he had a stomach virus, and treated him as such. After a few hours he started to feel better and wanted to eat. We let him eat, and he immediately started to vomit and complain of worsening pain. We called the pediatrician's office and were told that there was a stomach virus going around and he should feel better within a couple of days. Alex just kept getting worse though, and by Saturday night he was screaming in pain. We took him to the emergency room, where tests were run, a stomach virus was diagnosed, and they sent us home. By the next afternoon, Alex was still screaming in pain and continuing to vomit whatever liquids we gave him (trying to keep him hydrated).

On Sunday evening we took him back to the ER. I basically told them that THERE WAS SOMETHING WRONG WITH HIM THAT WAS NOT A STOMACH VIRUS and that we would not be leaving until they figured out what it was! They didn't like me, they thought I was a crazy pushy mother, but Alex was screaming in pain and they started doing all kinds of tests that they hadn't done the night before.

Because my family had, only 3 months earlier, attended the first WAGR Weekend in Virginia, we'd met the Luis family from New Jersey. While all of this was going on at the hospital, I had my mother contact Catherine Luis, whose daughter Irma has WAGR

Syndrome, and is 6 years older than Alex. What we were experiencing was familiar to Catherine, and she told my mother to have the physicians test for pancreatitis. Although there was absolutely no medical documentation connecting WAGR Syndrome to this disease, Irma had it, and Catherine knew that what Alex was experiencing she had seen before. To make a long story short, Alex was finally diagnosed with pancreatitis. He was admitted to the hospital, given IV pain medication, and kept off all food and water for the next 2 days. By the 3rd day, he was able to come off the pain medication and given clear liquids, then eventually low-fat foods.

Alex has had 8 hospitalizations for pancreatitis since that first one 7 years ago, and several episodes which were minor enough to allow him to stay at home. What we have learned in those 7 years is that some people with WAGR Syndrome are more likely to get pancreatitis than people who do not have the syndrome. Aside from Irma and Alex, we now know, through the support group, that there are other children with the syndrome who suffer from this disease as well. The connection between pancreatitis and WAGR Syndrome is one of the things being investigated as part of the current NIH study.

Alex has mixed hyperlipidemia-high cholesterol with high triglyc-

erides. He is on a low fat diet and takes medications to lower triglycerides. He also takes pancreatic enzymes with meals to aid in digestion. Parents should be aware that children with WAGR syndrome may be at risk for pancreatitis. Physicians should be made aware of this possibility, as there are some medications which can increase the risk of pancreatitis. Also, if you child becomes ill with the symptoms I described, and does not respond to the usual treatments, you may want to ask your physician to order blood work to screen for pancreatitis.

Written by: Karen Rose



Dear Families,

Greetings from Bethesda, Maryland! We are so grateful to the WAGR families all across the United States and all over the world who have enabled us to progress in our research over the past year. We appreciate your support, cooperation, and patience as we move along. We are fortunate to be working with such wonderful people.

To date, we have enrolled 43 patients with WAGR syndrome, hailing from 23 states in the US and 9 different countries. This is a great response, and we are still inviting new families to join!

It was a pleasure meeting many WAGR families this past July at the annual IWSA conference. In case we missed you, here is a summary of the presentation we gave on our recent research findings

After gathering medical records and collecting blood samples from families, we focused on investigating why some people with WAGR syndrome become severely overweight during childhood while others do not. Just as the deletion of *PAX6* causes aniridia and the deletion of

WT1 causes Wilms tumor, we hypothesized that the deletion of a gene called *brain-derived neurotrophic factor (BDNF)* is the cause of obesity in some patients. We found that this gene is missing in roughly two-thirds of WAGR patients, a fact that is explained by its close proximity to *WT1* and *PAX6* on chromosome 11. Previous research showed that *BDNF* deletion causes obesity in mice, and so we wanted to find out if it has the same effect on humans. After comparing patients with and without *BDNF* deletion for differences in body mass index (a measure of body size that takes into consideration a person's weight as well as his/her height), we confirmed our original hypothesis that *BDNF* deletion is associated with a higher risk of becoming overweight during childhood.

These findings were recently presented at two international scientific conferences (one was for pediatricians and the other was for endocrinologists) held in Toronto, Canada. The research was well-received by the scientific community, and we will be presenting these findings at two more meetings in October (one for obesity researchers and one for geneticists). As you can see, we really want to get the word out! We are also writing a paper for publication in a medical research journal.

We are currently in the process of planning the next phase of our study, in which we will conduct comprehensive evaluations at the NIH to more thoroughly explore the relationship between genotype (which genes are deleted) and phenotype (clinical symptoms). We will keep you updated and let you know when we can begin scheduling visits to the NIH!

Thanks again for everything!

Sincerely,
Rebecca Levinn, BA and
Joan Han, MD

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The IWSA is a
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IWSA 2007 Gorilla Awards

Gorilla Dad (to Arman) ~ Shaqir Arapi (Kosovo)

**Gorilla Mom (to Johanna) ~ Michelle Meyer
(Ohio, USA)**

Every single parent in our organization is a Gorilla parent every single day... but each year we select parents who have stood out to us in some way as someone who "embodies the spirit of the gorilla."

This award began at our first meeting in 2000. Seven parents met, for the first time, in Manassas, Virginia USA and Kelly Trout presented Catherine Luis a little gorilla as a token of love, appreciation and understanding of the way she'd fought for everything she needed to do in order to best care for her daughter, Irma. The Gorilla Award was born!

The concept of being "a gorilla parent" came from the animated Disney movie Tarzan. Tarzan's gorilla mother fought her peers, her family, her society and sometimes even her spouse, in order to do the best thing for her little human baby. He was different... and no one truly understood her complete and total adoration for this little creature... but she knew he was special and wonderful and well worth giving up anything she needed to... and fighting whoever she had to... in order to make sure he grew up healthy and strong and smart and safe.

She was his hero... and in many ways, he was hers. Her love for him strengthened her and she became more powerful than she'd ever been... to be strong for her baby.

Our gorilla parents are all that and so much more. While they're fighting all their own battles, doing it with grace and dignity and never letting go... they're also still supporting other parents as they go through their difficult days as well. They'll share their story if it will help another parent. They'll put down their own fears and grief to support another parent who's afraid or grieving. They're advocating for others as well as for their own child and themselves and sometimes they even have to pop the occasional doctor or service provider on the head to make things happen.

IWSA Officers & Board Members:

President - Catherine Luis
Vice President - Kim Pillow Williams
Secretary - Tammie Hefty
Treasurer - Becky Deas

Board Chairperson - Annie Prusakiewicz
Member - Rose Mallon
Member - Karen Rose
Member - Kim Pillow Williams

Michelle was present to receive her award at WAGR Weekend in Manassas. Shaqir received his award by mail.

Congratulations to the recipients of our 2007 Gorilla Awards, given with great love and respect, from International WAGR Syndrome Association.



Mark Your Calendars Today

The Prusakiewicz Family is asking everyone to join them for **WAGR Weekend 2008, July 11th - 13th** in Taylor, Michigan. (Just South of Detroit) Reservations can be made by calling the Ramada Inn (Taylor, MI) at (734) 283-2200 <http://www.showhotel.com/ramada/4818001/>

Ask for the WAGR Group rate

The discounted room rate is \$79.00 per night.

Friday Night - Meet & Greet, Mom's Night Out
Saturday - AM Group Photo, Morning Round Table Discussion, Afternoon Activity, PM Pizza Party, Dad's Night Out
Sunday - Family Social Time, PM Farewell Dinner

WAGR WEEKEND 2007



Tammie & Evie Hefty, WI

WAGR weekend was not an easy event for us to attend. In the past we've had other obligations on the same weekend, and this year we had obligations the surrounding weekends. But, with all that we've been through with the group in the past three years, we were determined to make the weekend happen for our family.

I had anticipated crying. A lot of crying. I was surprised, though, because I didn't have the shock of meeting everyone that I expected. Rather, I felt as though it was more of a reunion than a first meeting. I suppose with all the good days and bad days I've shared over the internet with everyone, you are more of a family to me than some of my other family.

Jeff and I both shared in the same joy, watching Evie play with other kids. I sat and watched Nicholas and Jenna do their stand up routine with the microphone and Evie and Hayden play with the jumbo legos and I felt like I was in heaven. Evie may not have noticed the difference, but I did. Almost more than seeing Evie play with other kids with WAGR, I enjoyed watching her play with the siblings of the kids with WAGR. Maddie, Hayden's

brother, adopted Evie like her own kid sister. The peace of knowing that Maddie accepted Evie the way she accepts her brother delighted my soul.

I appreciated that I didn't feel monitored. Sometimes, even with friends or family at home, when I want Evie to take a nap, people will press and say, "oh, she's fine." Okay, at that moment she's fine, but she doesn't always know when she needs a nap. At WAGR weekend, we understood, "hey, when the kid needs some time away, the kid needs time away!" Nobody said, "don't make her take a nap, how often does she get to see these people?" Wait, I take that back, Mr. Dreamy Dalton was really pressuring me to wake my daughter up from her nap so she could play, but I think he does that with all the girls.

Coming home from WAGR weekend was more of a culture shock than going to WAGR weekend. Suddenly, my skin felt a little tighter (maybe because I ate so well), and the air seemed a little heavier because my cyber family was once again only reachable through the keyboards and computer screens. But each one of them is here in my heart, and within a stroke of the keys.

Written by: Tammie Hefty



Kim Pillow, Irma Luis, Caroline Larson, Kelly Trout, Catherine Luis & Laura Trout

Our WAGR Weekend 2007 Memories

Caroline Larson: "I loved being there and meeting all the families and walking around taking pictures with my new camera. Talking with Dr Han and Rebecca seeing Carolyn again was so exciting. I can't wait to go back to the NIH. The first visit there was okay, but I was so nervous about what would happen. Now I know there's nothing to worry about, so I can relax and just enjoy it. Becky (Deas) and Irma (Luis) and I sat together and watched Ethan playing. He is so cute! I wonder how much I was like him when I was little. Taking pictures gave me a chance to talk to everyone. Like they say, photographs are worth a thousand words. And the memories - the memories will last a lifetime!"

Laura Trout: "I enjoyed talking with Michelle Meyer, she was so nice, and little Johanna is just adorable. Helen Lane and I talked about school - of course! She was really kind. Kristine Canavan (Liam's mom) and I were having a nice conversation when my mother spilled a coke all over the table and her keys! Maddy (Hayden's big sister) and I walked around the hotel together. She is so sweet; she had cut out stickers and gave them as presents to everyone. And I'll always remember Nathan (Tunis) speaking French!"

Kelly Trout: I think if I had my way, WAGR Weekends would be WAGR Weeks. The time we have together is always so short, and that makes each moment very precious. Every WW we've ever had has been magic, and this one was no exception. The presentations by Dr. Han and Ms. Ana Morales were terrific, of

SHARING MEMORIES

course. But as I watched them, I couldn't help feeling completely awed by the fact that these research projects are even happening. They are quite literally dreams come true. One of my favorite things is seeing friends who've come to previous WWs, and marveling at how much the kids have grown and changed. But one of the sweetest things about this and every WW is meeting families who are new to the group, and watching them realize that they and their child have found a safe and very loving haven. It's pure joy to know that next year, these "new" folks will be old friends, too.



The Dell Family, PA

We get very excited when July comes around because we know at the end of the month we will be seeing our wonderful WAGR family. I think the thing we like most about the weekend is that we just feel plain 'ole comfortable there. We have no one to explain anything too. Everyone there just

"gets it". If your kid is having a melt down, being a slob while eating, or doing the quirky things that they all do, it's okay. No one will judge you. No one will stare at you. No one will whisper to the person beside them about you. Being there just gives you a peaceful feeling. An "I'm not alone" feeling and boy do I love that feeling. Thank you everyone for a great weekend and giving me that feeling. I am proud to call all of you "My Family!"

Julie Dell, mom to Hayden 4 yrs old WAGR

This was the first WAGR weekend for me and my wife Karen. It was amazing, happy, sad and a little bit weird all rolled into one.

I got to meet Dr. Han in person. It had been just a few days before WAGR weekend that Dr. Han let me know that I didn't have a deletion. So, here we were at WAGR weekend. Neither of us had WAGR syndrome and we didn't have any kids with it either. All I had was a PAX6 mutation in an odd location. It didn't matter. Kim made us feel welcome and everyone was so nice.

The presentations were all excellent. From bio chips for screening to simpler sampling, the technology is really moving forward thanks to the smart people working in this field.

There were some sad moments too. It was sad when, at the end of the weekend, I realized that I hadn't mingled and gotten to know more of these wonderful people. My new family. This was a mistake that I don't plan

on repeating.

I thought this would be easy, but I have no idea how to put the whole thing into words.

P.S. The non-smoking location was so painful for me that after that weekend, I quit smoking. I just didn't want to feel that way again.

Written by: Walt Taninatz (NJ)



Shari, Amy & Jim Krantz, MD

It was great to meet everyone at WAGR Weekend. It was our (Amy, 12, and my husband Jim and I) first time meeting everyone and attending the "Weekend". I was overwhelmed with the kindness and kinship and am still recovering from the thought that I am not alone out there trying to figure things out.

It has been (and I am sure will continue to be) a long road and with a diagnosis finally of WAGR in Nov 06, I am still learning and researching.

While I was familiar with the information presented by Joan and Ana it was interesting to see in the graphic forms and to be in a room full of people

who has the same interests and dear children like me. I plan to visit the website and links to additional information as time permits, especially things dealing with medications for the ADHD piece. We have tried many different drugs with some success, but I am sure WAGR parents can help sort out some other info that we have not had access to.

My feedback about the "week-end" was that we attended Saturday (we live 45 miles from Manassas) and I was thrilled with the very warm, kind, and loving welcome of everyone. Just wish I had found you all a long time ago.

Written by: Shari Krantz

Friday was the Meet and Greet and boy, were we meeting and greeting! It was so wonderful to meet the new families and to see some old familiar faces that I hadn't seen as I was unable to attend the WAGR Weekend 2006 in Pennsylvania. Many of the siblings had grown at least a foot or two... and our children flooded in from every direction.

It's funny you know... all this connection we have on the internet. We feel so much like we know each other... and I was amazed at how many "strangers" walked up to that meeting room door and just by knowing so much about them and their children in writing, I knew them right away!

Before I knew it, it was time to wrap up the Meet and Greet and get ready for Moms' Night! We had a really good turn-out and had to wind up pushing several tables together so we could all see/hear each other. We had some old stories... and some new concerns... and just a lot of laughter and relaxation. We all needed it! We didn't actually wrap up completely until after 1a.m. with several of us dragging it out to the parking lot for a very slow good bye

session because it's just so darned hard to tear ourselves away from all this new family!

Ana Morales shared with us the OraGene kits and how simple they are to use. It's amazing! The whole kit is about the size of a cell phone! The basic kit is one where you just spit several times into this little cup, then screw the cap back on, shake it and send it back! If you have a child who cannot yet spit, then there's a kit containing five swabs and a small set of scissors. You swab the child's mouth with each of the five swabs, cut off the tips, drop them into the little capped container, screw on the lid, shake it and send it back.

No ice packs. No time limits because the kit can sit there for years before you have time to spit into it. No discomfort. No appointment, laboratory trip or time off from work. You just do this at home. Spit, shake and send. Cool.

Dr. Han brought some folks with her. She had a whole team up there and all were delightful and participated in her presentation. We're sorry to see Carolyn Menzie leave, but happy that she's being replaced by a wonderful young woman named Rebecca Levinn who is clearly enthusiastic about our families and the work at NIH.



Dr. Han's NIH Team

After lunch, we'd planned a sort of Question and Answer time, but it seemed that both presenters had been so thorough there weren't many questions at all and so many babies were cranky and tired and

parents were just exhausted so we decided to just break for the afternoon and get back together for dinner (Pizza Night.)

Then some families started leaving... I think having accomplished what they'd come for... and knowing they had a long drive back. So, we tried to throw together a group photo with the families who were remaining in the room and the presenters before they left town. We actually wound up doing TWO group photos because the other was already scheduled for right before dinner.

Dad's Night was small, as so many of our families had left already... but I think it was productive from what I gathered after... and these awesome Gorilla Dads took up the first few minutes of their meeting time to help me load my truck up and then cleared out the room for us at the end, so they had a working Gorilla Dad Night and for that I thank them all :)

Saying goodbye is never easy and while the dads were "going nuts" in their own space, a handful of moms (and almost moms - Marc's sissy Stephanie and Keisha) were doing our own thing in the breakfast area, connecting and saying goodbye as well. Tammie and Julie had taken water bottles full of leftover wine to their rooms to hang out together with the babies while the dads, so they were having their own little mini mom night - lol

I wish my vocabulary was larger because I feel like I'm using the same superlatives over and over in describing my own WAGR Weekend experience... but I can't find words in English large enough to say how much this event means to me. Perhaps my little pal Nathan Tunis can remind me of something bigger in French :)

Written by: Kim Pillow-Williams



Jeff, Clem, Brian & Tom

Nicholas, Hayden, Marc, Evie, Jenna, and Andrew respectively.

Next year, I'll know to plan to be out longer for the Gorilla Dads' get-together... and I'll also keep an eye out for the police.

Come Join Us

Email Listserv - Discussion Board

More than 150 people belong to the IWSA email discussion group. This listserv allows members to communicate with others who understand the joys and the challenges of caring for a person with WAGR syndrome. You may join the email listserv by following the link below:

<http://health.groups.yahoo.com/group/WAGR/>

The IWSA is a non-profit group, and is run entirely by volunteers. Membership is free. Funding comes from donations and grants. Because of our non-profit status as a public charity, we regret that we are unable to assist in fundraising for individuals or assist with medical expenses.

Forward newsletter submissions, stories, pictures or ideas to

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WAGR

We ate good, right?

We are Gorilla Radicals!

We are Guys Reminiscing-

by Tom Cox

The latter was certainly the case for the second annual Gorilla Dads' Night at WAGR Weekend 2007. I grossly underestimated the level of sharing we fellas were about to have when I sauntered down the hall through the hotel lobby into our meeting room at 9 PM Saturday night-- in fact, I told my wife I'd be back in just 45 minutes. Well, as we began to clean up just after midnight, two things had occurred: I had been proven wrong (not unusual) and the police had been summoned to the hotel (not for our group).

Clem Prusakiewicz, Brian Dell, Dean (Marc Martin's stepdad), Jeff Hefty, and I spent the bulk of the evening comparing WAGR war stories over a few adult beverages and snacks. Oh yes, Andrew Tunis from Canada popped down for an hour as well (not all the way from Canada-that would be too far to come!). I think we were all actually quite encouraged by our time together. You know, there are not too many people on the planet to whom you can say, "Radical right nephrectomy-how about you?" and have them understand completely what you mean and the arduous nature of your family's journey. We men might sometimes act like furniture, but Gorilla Dads are people too! It's good to know that we're not alone in loving

"The mission of the IWSA is to promote international knowledge and awareness of WAGR syndrome and its complications and treatments, to stimulate research and to reach out to those affected by WAGR syndrome in an effort to improve their lives"



Gorilla Mom's Night Out

University of Miami Research Study Update

In July, I had the pleasure of meeting many of you at the WAGR Weekend 2007. I had so much fun interacting with all of you. I was very impressed by the level of organization and care that Kim has put into this group. The hotel was beautiful and the food was delightful.

I also enjoyed meeting Dr. Han and her team, while learning about the exciting WAGR studies going on at NIH. Moreover, I was deeply touched by your warmth and your invitation (which I have seriously taken) to attend next year!

Working as a research genetic counselor is what I love the most. Coordinating genetic research studies has provided me with the opportunity to develop long term relationships with families, and I hope I can do the same with you. The WAGR study at the University of Miami started after a successful meeting when Dr. Fan shared his research goals with the WAGR Association board of directors.

Dr. Fan's research goals stem from the fact that autistic features are seen in about 25% of individuals with WAGR. Several studies have suggested linkage of 11p11.2-13 (the chromosomal WAGR region) with autism. Also, from the Human Genome Project we also know that this region includes about 70 genetic regions and 40 known genes. Knowing that there are so many genes in this area makes us want to learn more about this region, since we only know about 2 WAGR genes so far: PAX6 and WT1. Perhaps any of the other genes in this chromosomal region can explain more of the features associated with WAGR, including autism. With this information in mind, a research project was designed where a new tool, known as microarray, would be used to learn more about

autism spectrum disorders in children with WAGR, as well as other clinical problems associated with this condition.

Microarrays are about the size of a microscope slide. They test for gain or loss of genetic material in many different genes at once. Briefly put, microarrays can be thought of as an expanded version of the currently available routine chromosomal testing. This new technology detects genetic changes that would not have been identified by standard chromosome analysis.

Using microarrays, our study found very interesting results. A total of 25 WAGR families were recruited into the study. Results show that all 25 individuals with WAGR had different sized deletions on 11p13. The smallest deletion detected involved 30 genes, while the largest one involved 70 known genes. In addition to PAX6 and WT1, the deletions involved several genes known to be related to nervous system development and brain function: PRRG4, BDNF and SLC1A2. PRRG4 is involved in cell interactions and transport of substances known as neurotransmitters, or neuron messengers. BDNF plays a key role in neuronal development. SLC1A2 transports glutamate. Glutamate is an important amino acid for learning and memory. Therefore, our results suggest that, when there is not enough product from PRRG4, BDNF and SLC1A2, mental retardation and autism may occur.

There is still a lot of work to be done, such as further expansion of our analysis and additional samples that will add statistical power to our data. We would also like to correlate our findings with demographics such as age and gender, as well as inheritance of the deletion, which may provide additional insights about the possible mechanisms involved in development of autism in individuals with WAGR.

Completion of our work depends on obtaining additional samples. As I explained during my talk, we recently tested Oragene, a new method for DNA extraction. It is done from saliva instead of blood. The kit is the size of a cellular phone. By using it we can get plenty of DNA. No ice gel pack or phlebotomy services will be needed. The saliva collection can be done at home. The whole procedure should take no more than 30 minutes. The kit can stay at room temperature for years.

Since Oragene involves virtually no pain and a lot less effort to coordinate, I distributed many of these during the weekend, and we have already received some of the samples back. We look forward to receive the rest of the samples. However, if for some reason you changed your mind about participation, just let us know. Research participation is voluntary and you should not feel obligated to enroll.

The results derived from our research have already shed some light into some of the most difficult questions about WAGR syndrome. Our progress has been possible thanks to the generosity of many WAGR families. Their participation not only will increase knowledge about WAGR, but may eventually benefit all individuals with WAGR syndrome. Dr. Fan and I would also like to thank Dr. Han for her collaboration with our group.

We are all a great team. Research collaboration generates new possibilities, more curiosity, new partnerships, and new perspectives...and that's what it's all about!

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Pancreatitis in Children with WAGR Syndrome

Several children with WAGR syndrome suffer from a condition known as "chronic pancreatitis." Chronic pancreatitis is a serious problem. It can cause significant pain and in rare cases can even be life-threatening. Pancreatitis is not common in typical children, so it is important for parents of children with WAGR syndrome to be aware of the signs and symptoms and to alert their child's physician to the possibility.

There are two reasons why children with WAGR syndrome may be susceptible to pancreatitis. First, the PAX6 gene is important not only in the development of the eye, but also in the development of the pancreas as well. So some people with aniridia may also have defects in the anatomy or function of the pancreas. In addition, many of our children have high levels of fatty molecules in the blood, called "lipids." This condition is called "hyperlipidemia," or "hypertriglyceridemia," (having high levels of triglycerides, a particular type of fatty molecule). It is thought that having high levels of fats in the blood increases the risk of pancreatitis.

Pancreatitis is inflammation of the pancreas, which is a large gland behind the stomach. The pancreas secretes digestive enzymes into the small intestines through a small tube called the pancreatic duct. These enzymes help digest fats, proteins, and carbohydrates in food. The pancreas also releases insulin and glucagon into the bloodstream. These are hormones that help the body use the glucose it takes from food for energy.

Typically these digestive enzymes do not become active until they reach the small intestine, where they begin digesting food. If these enzymes become active while still inside the pancreas, they start "digesting" the pancreas itself.

Acute pancreatitis occurs suddenly and lasts for a short period of time. Chronic pancreatitis does not resolve and can result in a slow destruction of the pancreas itself. Either type of pancreatitis can cause serious complications including bleeding, tissue damage and infection. In extreme cases there can also be damage to other organs.

Signs of acute pancreatitis can include:

Swollen and tender abdomen, Nausea, Vomiting, Fever, Rapid pulse, Anxiety and Restlessness or irritability

During an acute attack, the blood contains at least three times more amylase and lipase than normal. Amylase and lipase are digestive enzymes formed in the pancreas. A doctor can order a blood test to diagnose acute pancreatitis.

In chronic pancreatitis, patients may have abdominal pain. The pain may get worse when eating or drinking. The pain may spread to the back or become constant and disabling. Other symptoms may include nausea, vomiting, weight loss and fatty stools.

Treatment for pancreatitis might include pain relief, placing a patient on an IV or all liquid diet following an acute attack, and then gradually moving to a diet high in carbohydrates and low in fat. Antibiotics may be prescribed. A doctor may also prescribe pancreatic enzymes to be taken with meals if it is determined that the pancreas is not secreting enough enzymes on its own.

Some children with WAGR syndrome appear to have an altered sense of pain. Because of this, your child's physician may wish to assess each event of nausea and vomiting, especially if the child appears to have constant recurrent "viral illnesses" that do not affect other family members. We encourage you to speak with your child's physician about the incidence of pancreatitis in children with WAGR syndrome.

We hope that our ongoing research projects will help us to understand more about why our children are sometimes affected by pancreatitis, and what we can do to help.

You can read more about pancreatitis on the NIH/NIDDK web site: <http://digestive.niddk.nih.gov/ddiseases/pubs/pancreatitis/>

And here:

http://www.emedicinehealth.com/pancreatitis/article_em.htm

From IWSA Board of Directors

The Board of Directors met on Friday, July 20. Those present were Annie Prusakiewicz, Rose Mallon, Kim Pillow Williams, and Karen Rose (by speakerphone.)

The Directors discussed organization finances and a report of all expenses for the last two years. It was decided that we would purchase software used by non-profit organizations to help us organize our financial information better so that we can protect our non-profit status with the IRS. Soon we hope to have the capability to project our expenses into the coming year, two years, five years to even better serve our purpose and meet our goals and objectives for the organization.

We had discussion about several legal issues including insurance and the use of our new logo. We made the decision to redesign our brochure and discussed some ideas. We decided to print the newsletter, in color, twice each calendar year. We also decided to put the Parent Welcome Packet onto a CD that could be easily mailed out to each family to reduce printing and postage costs. Also this method makes it easier for a family to share the CD with their child's physicians.

We discussed and considered sending a representative from IWSA to Genetic Alliance conference next summer. We discussed

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

our current research projects, the progress and status of each and consideration of creating a MedQuest 2007/2008 so that as an organization we might update our own statistics about the conditions associated with WAGR syndrome. This was so helpful in the past, allowed us to collaborate with physicians on article publication and it was noted that there will never come a time when EVERY family will participate in any research project so we maintain responsibility to have that information available for families and physicians.

Further decisions will be made about WAGR Weekend 2008 at our October board meeting and a form will be created to be used by families requesting sponsorship for the event. This form will be in the Spring 2008 newsletter along with more WAGR Weekend detailed information.

We had a great deal of discussion on how we, as leaders, could all be more effective for our organization and how we can support each other in the performance of our duties. We also discussed the merging of information from Reaching Out web site into IWSA web site to have a more united and clear presentation via internet.

We discussed the need for an annual report so that we are able to more accurately track our activities, especially important when writing grants for funding.

At our next meeting we will be electing/re-electing the leaders of our organization.

Anyone wishing to share time, talent, energy, organizational skills or business experience with the leaders of International WAGR Syndrome Association, please contact Kim Pillow Williams at: towandakim@yahoo.com

We are all volunteers and more hands will lighten the load!



**Nicholas Prusakiewicz,
11 yrs old**

Fall is in the air here in Michigan. Nicholas is back at school. He was very excited to start school this year because he was selected for a very important job - the safety patrol. He is positioned at the front doors. He loves to open and close doors so this is a perfect spot for him.

Nicholas is in the 5th grade this year. He spends most of his day in a self contained Cognitively Impaired classroom. This year he only has 4 other classmates in his room. He will continue to work on his core subjects in the resource room. He will also have the opportunity to be with his regular 5th grade class for gym, music and art.

This year we will start the process of transitioning Nicholas to Middle school. I am stressing just a little trying to search for a program that is appropriate for him.

Nicholas is holding his own medically. The cataract in his left eye has gotten a little more dense. But his ophthalmologist and we agree that surgery is not necessary at this time. Nicholas still has good functional vision. We hope to hold off on any eye surgeries for as long as possible.

Nicholas will begin his fall horseback riding program this month. He really enjoys riding horses. His most favorite thing to do with his horse is trot. I also enjoy watching him ride.

Nicholas turns 11 years old this month. I can't believe that my "baby" is eleven years old already. It just doesn't seem possible. Nicholas is growing into a wonderful young man. He has a great sense of humor, is very caring and brings so much joy to our lives.

We had a fantastic time out in Virginia this summer. It was so nice to meet all our new families as well as getting to visit with all our familiar friends. For me, the weekend ended too quickly and it was very hard saying good bye.

We want to extend a special invitation to everyone. We hope that you will consider joining us in Michigan for WAGR Weekend 2008 next July 11-13th. If you have any questions, concerns or need assistance, please contact me at TheMooZoo@aol.com or at (313) 381-4302.

Take care everyone,

**Annie Prusakiewicz,
Michigan**