



# WINGS

WAGR Information, News, Gorilla Stories

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

www.wagr.org  
Spring/Summer 2008

## From the Chair...

Dear Families and Friends,

I am so happy to have the opportunity to check in with everyone again. I hope that all is well with you and your family.

The Board of Directors unanimously appointed two new board members in February. Both members bring a wealth of leadership and professional skills to the organization.

Thomas M.Cox (Jenna's dad) has been a member of our group for a number of years. He lives with his family in Pennsylvania. He and his family have attended a number of WAGR Weekends over the years and even hosted this most celebrated event back in the summer of 2006.

Tom says "Being a parent of a WAGR child myself I would like to use my experience to minister to the needs of other families in similar situations. I believe in the mission of the IWSA and have long been hoping for the opportunity to help further such a cause."

Shari Krantz (Amy's Mom)

amazingly found our group last summer right before WAGR Weekend. Shari lives with her family in Maryland. We admire Shari's energy and the new perspectives she has brought into the online discussion group.

Welcome again new board members, Tom & Shari.

The Board of Directors also appointed Catherine Milian (Enzo's mom) to the position of IWSA Fundraising Coordinator. Cathy will be working with the officers and board members to develop and implement fundraising ideas to generate revenue for the organization. Catherine has been actively involved with charitable activities for various organizations. We are very happy to have her on our team.

Julie Dell (Hayden's mom) was appointed to the position of Assistant Secretary. Julie resides with her family in Pennsylvania. She is also our WAGR Historian and does a tremendous job capturing all our WAGR memories.

Just a reminder ~ our private discussion group is a comfortable place to share your child's story and meet other families. If you are not already a member we welcome you to join.

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

In the near future we will be launching a Wilms' Tumor / Renal Function Survey and ask that each member consider participating. We will keep you posted when this project begins.

The NICHD/NIH WAGR Research Presentation is available via e-mail or on disc for families upon request. Please feel free to contact us for your copy. [Reachingout@wagr.org](mailto:Reachingout@wagr.org).

Catherine Luis has done an amazing job maintaining the [wagr.org](http://wagr.org) web site. If you have not visited it lately, please stop by to see all the latest information and the other great things the IWSA has to offer.

Con't pg. 12 ~ see Chair

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## Gorilla Stories

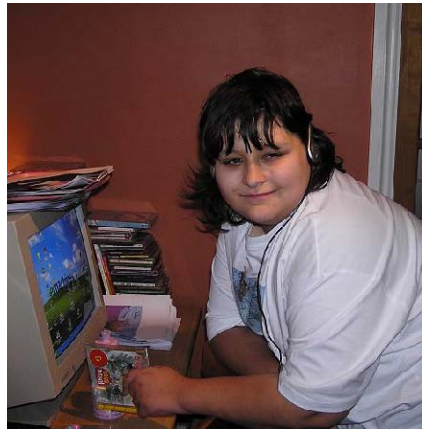
Just the thought of moving house with a child with an autistic spectrum disorder would be enough to bring you out in a cold sweat. To bid farewell to everything that for 12 years had become her home environment. Every room, colour and smell. The cupboards, the carpets, the garden, the doors, everything would be totally different. There would be no gradual introduction, no getting used to it, just wam - bang - wollop. One day she left for school from the old house and returned from school to a different one !

An endless nightmare of "Abbey go home!", "Abbey go home!", "When can Abbey go home?". Tantrums?, head banging? screaming?, pinching? Sound likely? Sound familiar? It's certainly what we were expecting.

In fact, however, there was absolutely nothing! Not a single murmur. From the day we moved we've hardly had mention of the "old house" and this from somebody who HAS to follow a routine. No matter how much we try and educate our selves in the facts and theories, Abbey just continues to take us by surprise and we were able to settle into a new routine without any bother. That was two years ago now.

Waking up on a Saturday morning is always a little easier going than it is on a work day or on a school day. At least you feel like there's no pressure. That is, until Abbey starts on her first run through of the day's itinerary.

"Toast first Dad-say it Daddy -  
"Toast first"  
"Then it's ladies - yes ?"  
"Then it's dinner"  
"Then it's Nanny's Office"  
"Alarm on"  
"Shopping at savacentre"  
"MacDonalds"  
"Nanny's House"  
"Then it's Nanny bring me home -  
yes ?"  
"Say it Daddy, come on, .....Toast  
first ....."



The first of many times we have to run through the day's schedule.

The "ladies", (second on the itinerary), is in fact, a snippet of respite care that is afforded us by the local social welfare department. They arrange for a couple of part time carers to come and collect Abbey from the house on a Saturday Morning. They generally take her where she wants to go which is usually to one of the local parks or to her favourite shop on the planet, the Disney Store. She's gone for exactly 3 hours, so provided we can muster the energy, Jackie and I sometimes use this time to go to the DIY store or the garden centre. Or, it's even been known for us to just go for a walk in the local country side! Leigh, our son, is now nearly 13 and old enough to be left alone in the house under strict instruction not to answer the door. It's a good job really, because he's at an age where a trip to the DIY store with Leigh in tow is not a very pleasant experience. For someone with no disability, it's amazing how soon his "legs start to hurt", "he get's thirsty and HAS to get a drink", and then has to question us over why we had to come out in the first place!

"Nanny's Office" thru "Nanny's house" (Itinerary 4 thru 8) is also a tiny bit of respite in that Jackie's mom takes Abbey from the office where she runs her small business back to her house for tea and to watch her DVD's in Nanny's kitchen. Abbey carries a small collection of DVD's around with her in a small holdall along with an arm

full of A4 (letter sized) pictures which are both her "friends" and her full time hobby. Sorted into groups by theme and sometimes each one different from the next by just the slightest thing, she knows them all individually and seems to just love the process of reproducing them from her computer printer.

It costs us a considerable amount in ink-refills for the Epson she uses and we learned very quickly to carry a matching spare printer in case of a mechanical failure. For Abbey, not to be able to print her pictures is like the world has come to an end. Except her instinctive reaction to a printer failure is one of anger, as it is in fact, when anything doesn't go her way. But then there is seemingly no way we can explain to her in a way she can comprehend, or at least is prepared to accept, that PC World is not open 24 hours a day, that we can't afford to simply replace it on a whim, or that the bulk delivery of ink on order is stuck in the mail, or even that it's 10:30 at night and time to stop and go to bed ! So we get beaten up.

But, when we've got plenty of ink in stock; the printer's working; the DVD player on the computer is ok; her DVD's are not too scratched; there's a musical instrument at hand; Jackie and I are where we're supposed to be in the kitchen; when Leigh's upstairs in his bedroom and there are no visitors expected, then everything in her world is fine. She's happy. And when Abbey's happy, she's very funny. She loves to make people laugh and finds achieving it hilarious herself. Once you've started, you keep getting reminded to laugh in the right places, which in itself is quite funny (at first).

Absorbed by her music, she likes to play extracts from DvD sound tracks which she has me produce for her on the computer. I make mp3 files so she can also carry them on her portable mp3 player . She will spend most evenings printing stills captured from her dvd collection while listening to something completely unrelated. She

loves classical music and sometimes likes to imitate famous opera singers, which we find very amusing but I expect not so much fun for the adjoining neighbours, and as if guided by some fashion sense she's now back into the spice girls (currently touring England again).

Leigh on the other hand, is more into guitar music or rock music, all of which he sources from the Internet. Accessed through a big bank of equipment in his bedroom, this is Leigh's human interface into a virtual reality. Whether it be via Xbox, PS, PSP, Wii, PC or DS, Leigh is more comfortable in his alternative world, the real world being just too boring for a 13 year old nowadays. If I'm honest, if he hadn't become an expert in all this stuff, I don't think he'd be able to communicate with any of his friends, as they're all seemingly doing the same. He's quickly growing up now though and becoming sensitive to how different things are for us as a family. But as long as we keep our reactions to Abbey's outbursts in proportion, he's able to tolerate the disruption it causes and not get too disturbed by it.

I regularly express my gratitude to God that we haven't had the more frightening of the medical conditions to contend with. Although that's not to say that 14 years of kidney scans is not harrowing enough. The threat will never go away I suppose. But her two main areas of concern, health wise, are one, her eye's and the terrible glaucoma she suffers, and two, her weight. I suppose that for various reasons, one being that she's now totally blind in her one eye, she really doesn't get to exercise anywhere near enough. This is on top of what we're convinced is a



problem connected with the syndrome, she now weighs more than me !

She also suffers regular urine infections, which if they persist will, I'm sure be investigated further. We've just finished a course of antibiotics which was the second prescribed in the last 6 months.

As I write, we're recovering from another small trauma, thankfully not connected with WAGR, but frightening never the less. As anything is I suppose when a child can't communicate the exact problem to you. She started to become feverish and very docile which is so unlike her at any time, and it took 3 doctor's home visits to diagnose the connection to a faint rash on her leg, but one that was worsening by the day. By the third day, she was running a temperature of approximately 102 and her leg was red and sore and swollen. She was (I'd say) verging on falling unconscious when they decided to call for an ambulance. However, whether it be through a rush of adrenaline or sheer fear of hospitals, despite 3 paramedics, two sets of grandparents and Jackie and I, we could not get her to leave the house ! The doctor returned and seeing her more "with it" said we could try dealing with the suspected infection by normal means and perhaps could avoid the trip to hospital and everything that would entail. Anyway, she's well on the mend now, all except her leg which is now looking like it's going to turn into a big blister.

Abbey still attends a special school for children with multiple and some profound learning difficulties of which Jackie is on the board of governors. In fact she is the deputy chair of the board and also a trustee of a registered charity, "Friends of Old Park School" (FOOPS) which has so far managed to raise thousands of pounds to help provide equipment for the children who attend the school. One of their fund raising ideas was for me to collaborate with the music teacher and write, record and produce a "school Christmas rap". I'm pleased to say, it worked out quite well, and the children

performed it as a finale to their Christmas play 2007. We sold nearly 100 copies of the rap on CD!

I'm still at Jaguar Cars for the time being, working in the manufacturing IT department looking after the systems that control and monitor the robots etc.. As you may know the company is being sold by Ford along with our sister company Land Rover. You can imagine that it leaves us feeling a little uncertain about the future. But then as I'm reminded, I don't think anyone has that "job security" that was once seemingly common place. Not in this day and age.

Well I think that's about all from me. We might not have contributed to the WAGR forum just lately, but we haven't forgotten you and still treasure the knowledge that you're all there and ready to help when we need it. (is that too selfish of me ?) We still get the emails in summary form and I try and catch up when I can. Hello to all the new members of the group, and a special hello to all those who we had the wonderful privilege to meet in person at the WAGR weekend in Manassas. A trip we will never ever forget for as long as we live. If only we could even dare imagine Abbey on a plane again for 10 hours, we'd do it again at the drop of a hat.

Neil, Jackie, Abbey & Leigh  
Sprason.  
Stourbridge, England.

**Forward newsletter  
submissions, stories,  
pictures or ideas to:**

**Annie Prusakiewicz  
P.O. Box 392  
Allen Park, MI 48101  
E-mail:  
TheMooZoo@aol.com  
(Please note our new  
address)**



## A Silver Lining

**M**y big brother Christopher has WAGR syndrome, he's protective, annoying and inspiring just like every big brother should be.

My name is Jenny, I'm 29, and Chris is nearly 31. My parents swear I wasn't an accident, as there is only 15 months between my big brother and I. I take my hat off to my parents having another child so soon after having their world turned upside down, and with an uncertain future with Chris and the never ending and surprising world of WAGR. In the master plan of things, apparently I was born to look after Chris, for him to have someone to follow and learn from. And that's exactly what he did when we grew up.....that is of course until my younger brother Michael turned up.

I don't really remember much of Chris and I growing up, although apparently we were inseparable. Even when he had a Wilms' tumor at the age of 5, I look back at pictures of him with no hair and wonder when I ever knew he was 'different' or that our family wasn't normal, or that something was wrong.

Over the years I have realized he is my normality, and every family is different. There was never a defining moment. We were always visiting him in hospital in Great Ormond Street in London, I just remember playing on the rocking horse, and sitting in waiting rooms a lot, and the smell, that horrible 'clean' hospital smell. It wasn't till I got to my teens, and I was trying to be 'cool' that I realized Christopher was not 'cool'. If he sat in the front of the car he always wanted his daft, loud, silly music on...nothing from the charts that I wanted to play. If he sat in the back of the car he pretend to drive with his Frisbee as the steering wheel.....dribble running down his face as he made the sound of the engine, the window slightly open as he was using the window handle as the gears! This same Frisbee would follow us to the shops as he 'pretended to drive around'...making the same loud noise. As his 13 year old sister this

was highly embarrassing. Everyone would stare!

Even on holiday...you couldn't miss our family.....Chris would be by the side of the pool on a quiet summers day, at the top of his voice shouting 'ready, steady, 3, 2, 1...Go! before he dive bombed in!!! My friends on that holiday knew me as 'oh you're the girl with the disabled brother'

But as you get older you realize, this is your family, this is who he is, you grow out of being embarrassed, and in fact I went through a stage of being really protective of disabled people. If any of my friends laughed or poked fun at someone who was like Chris...I would let them say their piece, do their impression... and just wait till they had finished, then belittle them.... and explain how stupid they were for being ignorant! I remember when I was 16, my parents had gone out one evening and I was left to baby sit Chris. It was one of the most scariest evenings of my life. He had recently fallen over and broken his hip, his hip was now pinned back in place. In the middle of the evening when I was downstairs watching TV and he was in bed, I suddenly heard a shriek from his bedroom, I have never in my life heard someone cry and scream in so much pain. I couldn't make him stop, I couldn't make it better. I called my parents and sat on the stairs, crying with my head in my hands until they got home. His muscles had gone into spasm, and were rejecting the pins inside his hip, one pin was actually poking through the skin and my dad managed to pull it out. Chris went back to hospital, as they had infect messed up the procedure, this is not the first time Chris has been let down by hospitals and his health... but that's another story.

Chris was my inspiration at University, as I researched his syndrome fully, as a Fine Art student I produced an exhibition on Chromosome defect and sight. A few years later at film school, my first documentary was on the friendship between Chris and my dad. Last year, and at the age of 30 Chris had his Kidney transplant, my dad was the live donor. As my mum was looking after my dad, I went down with Chris to the operating theatre,

to keep him calm whilst he was being anesthetized. I held the tube over his nose as we talked about what he was going to dream about...I actually thought it might be the last time I was ever going to see him. As he drifted off to sleep I burst into tears.

What I forgot was that Chris had been in and out of hospitals his whole life...he is so strong and knows exactly what's going on, it was me that was rubbish at holding things together, I was there also when he came round too....I knew he was fine when he told me to go away!

I suppose as adults, as a normal brother and sister, at times we can love to hate each other, with him having autism, he gets bored of me sometimes, and I still get annoyed that I have to compete with him in order to talk to my parents if we are in the same room. And one Christmas I was teasing him about whose new CD we were going to listen to, mine or his, when he hit me, but as I hit him back, my mum walked in...she couldn't believe I hit him back...but 'he hit me' I said...'of course I'm going to hit him back!' I felt so guilty as for the rest of the day he said he was sorry and would share his music with me! I wouldn't change him for the world! He is the sweetest and at times funniest person, of course he is annoying and loud, and if you're not in the right frame of mind to listen and answer his repetitive questions he can drive you mad! But he's my big brother and I love him for who he is.



Chris & Jenny - England

**You are cordially invited to attend  
WAGR Weekend 2008 - Taylor, Michigan  
July 11- 13, 2008**

**Ramada Inn of Taylor**

20777 Eureka Rd.  
Taylor, MI 48180

Phone: (734) 283 -2200 Fax: (734) 284-0967

Ask for the WAGR Group Rate (\$79.00/night)  
Includes Continental Breakfast each morning  
<http://www.showhotel.com/ramada/4818001/>

E-mail reservations may be made by contacting  
**Timothy R. Nafso, MBA - General Manager**  
E-mail: [RamadaDownriver@aol.com](mailto:RamadaDownriver@aol.com)

Tentative Itinerary:

**Friday, July 11, 2008**

6-8 PM - Meet & Greet (Cookies, Coffee, Tea, Soda & water served)  
8:30 PM -? Mom's W(h)ine & cheese night // This is for Mom's ONLY!

**Saturday, July 12, 2008**

10 AM - noon - Officer Presentation, Information Exchange  
Noon - 1 PM - All U Can Eat Deli Lunch Includes: Soup, Fried Chicken, desert, rolls, potato & Macaroni salad, Ham, turkey, & roast beef, condiments / hot dogs & mac n cheese available for children  
2-4 PM Family Fun - Bowling Bonanza at Taylor Lanes  
6-8 PM - Pizza Party Includes: Pizza, breadsticks, fresh garden salad & soda  
8:30 PM -? Dad's Night Out // This is for Dad's ONLY!

**Sunday, July 13, 2008**

Morning free time - Take time to mingle with parents and just relax  
3-5 PM Farewell dinner, wrap up, and good-byes Includes: Baked herb chicken, vegetarian lasagna, Caesar salad, fruit salad, au gratin potatoes, green beans, & dessert

**All catered meals and events listed above are free!  
However, the IWSA needs an accurate count of how many  
adults and children will be attending each meal by June  
15, 2008.**

To confirm number of adults and children in your group simply visit [www.wagr.org](http://www.wagr.org) to submit the information online, e-mail the information to Annie Prusakiewicz at [TheMooZoo@aol.com](mailto:TheMooZoo@aol.com) or call 313-381-4302.

Family assistance grants may also be available by contacting Annie Prusakiewicz at [TheMooZoo@aol.com](mailto:TheMooZoo@aol.com) or 313-381-4302 for an application. **Deadline for submission is May 31, 2008.** Priority will be given to families who have never attended a WAGR Weekend.

Additional Information:

The 2008 Meijer Taylor Summer Festival – the Downriver area's biggest and best festival – is scheduled for July 10 to 13 at beautiful Heritage Park, 12111 Pardee Road. The spectacular Masco Fireworks are scheduled for Friday, July 11. The festival features four days of carnival rides, games, special events, entertainment, food, beverages and fun for the whole family. Special events include some great concerts featuring top performers. For more info see <http://www.cityoftaylor.com/>

Families may also want to stop in and visit The Henry Ford - the history destination that brings the American Experience to life. Visit <http://www.hfmgv.org/>

# WE NEED YOU

The IWSA is currently searching to fill the open Vice President position vacated by Kimberly Pillow Williams late last fall.

There aren't any prerequisites. All of our board members and officers do what we do out of the love for our children. Many of us never had any official training or schooling to do what we do. It's more like on the job training, and we continue to learn as we go.

So if you think you might be in the market for a non paying, non glorified, but very rewarding position this may be the perfect job for you.

Vice President duties might include assistance with the following:

- \* Welcoming new families & inviting them into the e-group
- \*Following up on families well being (e.g. writing to families to see if there is anything we can do to help them. etc.)
- \*Connecting two families together when asked, if possible
- \*Helping to find resources for families if necessary/requested
- \*Helping to find resources for our web site and our organization
- \*Helping to communicate with other organizations on behalf of our families
- \*Communicating with other organizations on behalf of our families
- \*Assisting with fundraising ideas
- \*Communicating our organizational activities to the e-group on a regular basis

We are willing to mentor the right candidate, one with a great attitude that is able to go the extra mile for our families and help us fulfill the mission of the International WAGR Syndrome Association.

Time requirement varies.

For more information, please contact Annie Prusakiewicz at [TheMooZoo@aol.com](mailto:TheMooZoo@aol.com) or at (313) 381-4302.

## NIH Research Study Update & Information

### What is the NIH?

"The National Institutes of Health (NIH) is the primary Federal agency for conducting and supporting medical research. Helping to lead the way toward important medical discoveries that improve people's health and save lives, NIH scientists investigate ways to prevent disease as well as the causes, treatments, and even cures for common and rare diseases. Composed of 27 Institutes and Centers, the NIH provides leadership and financial support to researchers in every state and throughout the world." Visit the website for more information: <http://www.nih.gov/about/NIHOverview.html>.

### Where is the NIH?

It is located in Bethesda, Maryland, near the nations capital, Washington, D.C.

### Who are Dr. Joan Han and Rebecca Levinn, and what are their credentials?

Joan Han, MD is the principal investigator on the WAGR syndrome study. She is a board-certified pediatric endocrinologist. Dr. Han attended 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.

Rebecca Levinn, BA is a research assistant working with Dr. Han as the coordinator of the WAGR syndrome study. She graduated from Hamilton College in upstate NY, with a degree in chemistry.

### What is the purpose of the NIH/WAGR syndrome study?

We want to learn more about how genotypes (the specific genes deleted) are associated with phenotypes (the clinical symptoms present), since every individual with WAGR is unique. By learning how specific genes affect health,

we aim to improve the medical care of patients with WAGR syndrome by custom-tailoring surveillance and treatment recommendations to each patient's specific genotype.

### Who is eligible to be in this study?

Anyone who has had genetic testing showing a deletion on chromosome 11, or if the child has aniridia plus at least one other symptom typical of WAGR syndrome (Wilms tumor, genital/urinary abnormalities, and/or developmental delay).

### What are "Phase I" and "Phase II"?

Phase I is an outpatient study and consists of a fasting blood draw and the collection of medical records and a complete medical history for the child. One or both parents may choose to participate by having their blood drawn as well, but this is entirely optional.

Phase II is a week-long inpatient visit to the NIH, where we will conduct a comprehensive evaluation to explore the relationship between gene deletions and clinical symptoms. The studies will include blood and urine tests, radiology imaging, and examination by doctors in multiple subspecialties. This phase is about to launch and we will let you know as soon as we start scheduling visits to the NIH.

### What are the benefits of participating in the NIH/WAGR syndrome study?

Participation in this project may not result in direct benefit to your child, but the information that we learn from your child's participation will contribute to our knowledge about WAGR syndrome. Results of all clinical tests will be sent to you. Also, if any abnormalities are found during our studies, we will inform you and your child's doctor and provide recommendations for treatment and follow-up.

### What do we have to do to be in this study?

For both Phase I and Phase II of the study, we require that you

complete the various consent forms and informational sheets for your child. We ask that your child's doctors send us copies of any past medical records (we pay for all shipping costs). We also request a comprehensive medical history. This is just a series of questions, completed by filling out a form by writing, typing, or speaking on the phone with us.

For Phase I (the outpatient blood draw and collection of medical history/records), we provide all the supplies to collect and ship the blood samples as well as the forms and envelopes to give to your child's doctor to request medical records. The blood draw can be arranged in your home (we can send a phlebotomist to your house) or at your child's doctor's office (we will contact the doctor to assist with the arrangements). For young children (age <5 years) and for anyone uncomfortable with blood draws, we recommend performing the blood draw during a previously-scheduled blood draw appointment at your child's regular physician's office, or when the child has an exam under anesthesia.

For Phase II (the one-week inpatient visit to the NIH), our clinical center is open year-round, and we will schedule a visit during a week that is most convenient for your family. Prior to your visit, we will provide a detailed schedule of all the tests to be performed and all the subspecialty doctors who will be seeing your child, so that we can answer any questions or concerns you may have. Our goal is to custom-tailor your child's visit to best suit his or her needs.

### English is not my first language. Are interpreters available to help me?

The NIH has interpreters available for many languages, including sign-language. In-person services are provided by people trained in medical terminology and interpretation. For Phase II, please let us know in advance if you require language accommodations and we will make arrangements during your stay at the NIH.



**If we participate in Phase I, do we have to participate in Phase II?**

No, participation and continuation in this study is optional at all times, and you may withdraw at any point. However, any blood samples or medical information that we have may still be used for our research.

**Are there any costs for participating in Phase I?**

No, the NIH covers all costs for the blood draw and any mailing costs for sending medical records. We also make long-distance phone calls so that you do not have to pay for this (i.e. we will call you if we conduct the medical history questionnaire over the phone).

**We did not enroll in Phase I - can we now enroll in Phase II?**

We prefer to collect blood samples in advance of your visit to the NIH, so that we can have lab and genetics results available prior to the comprehensive clinical evaluation. In the case that having blood drawn before the Phase II visit is not feasible, we can wait until you are here. However, we request that all of your child's medical records are forwarded to us before your visit to the NIH.

**My child really hates blood draws. Is there anything we can do for that?**

To avoid an extra blood draw, we do our best to schedule this part of Phase I during a previously-scheduled appointment, such as a doctor's visit when blood is already being drawn, or an eye exam under anesthesia. We also use a numbing cream on the skin to lessen the discomfort.

**Can we enroll by sending just our child's blood?**

No, NIH regulations require that everyone signs a consent form before participating in any aspect of a study. Once enrolled however, only the child's blood is required; sending blood samples from the parents is entirely optional.

**What tests are done on the blood samples?**

For Phase I, we perform over twenty tests that include genetic

studies, cell counts, chemistries, lipid analyses, and hormone levels. For Phase II, we perform additional studies, including urine analyses and specific tests for pancreas function and diabetes screening.

**What happens if a problem shows up?**

You will receive a copy of all the clinical test results. If any of the results are abnormal, we will inform you and your child's doctor and provide recommendations for treatment and follow-up. During your child's inpatient visit to the NIH (Phase II), if your child becomes ill or is found to have an abnormality that is serious enough to require immediate medical or surgical attention, we will ensure that your child receives the appropriate care. The NIH is equipped with a state-of-the-art surgical suite and intensive care unit, and emergency transportation to a different facility is available in case a patient requires additional care outside of the NIH.

**If we choose to participate in Phase II is there financial assistance for travel expenses? For whom?**

Yes. The NIH will cover travel expenses within the (continental) United States for the patient and one parent or guardian. This includes airline tickets as well as ground transportation. There is a free shuttle available between the airport and the NIH campus in Bethesda.

**Where do families stay while visiting the NIH?**

The Children's Inn is located a stone's throw away from the Clinical Center, and serves as a "place like home" for patients and their families while participating in studies at the NIH. There is no cost to families staying at the Inn, and the entire family (parents, siblings, grandparents, etc) is welcome to stay. Their website contains more information: <http://www.childrensinn.org/site/c.kk11KIMXlvF/b.2001915/>

**Will NIH provide a letter for me to apply for a travel visa?**

Yes.

**I live overseas and cannot travel to the US is there any way we can participate in Phase II?**

The NIH can only provide flight assistance for travel within the United States. However, we can try to put you in touch with social service organizations that may be able to assist you.

**Will there be funding to assist families from overseas to get to the US?**

No, unfortunately we can only provide financial assistance for travel within the United States. Once in the US however, we are able to cover transportation costs of the patient and one accompanying adult to the NIH.

**Who should I call or e-mail if I have other questions?**

Please feel free to call or write either Rebecca or Joan if you have any questions; we'd be happy to talk with you!

Joan: (301) 435-7820;

[hanjo@mail.nih.gov](mailto:hanjo@mail.nih.gov)

Rebecca: (301) 402-6762;

[levinnre@mail.nih.gov](mailto:levinnre@mail.nih.gov)

***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.**

## ASK THE DOCTOR

The following answers were provided by Brian P. Brooks, MD, PhD --Diplomate, American Board of Ophthalmology & American Board of Medical Genetics

Dr. Brooks is a staff clinician at the National Eye Institute.  
E-mail address: brooksb@mail.nih.gov

**Q: In the past, "artificial iris" contact lenses (lenses with an iris painted on) were routinely prescribed for patients with aniridia. It was felt that these contact lenses would decrease light coming into the eye, protecting it from glare and improving both vision and cosmetic appearance. More recently though, routine use of contact lenses for people with aniridia has been discouraged. Why is this? (increased risk of exacerbating aniridic keratopathy?) Are some contact lenses acceptable for use by people with aniridia?**

A: You are correct. The cornea (clear window in the front of the eye) gets all its oxygen from the tear film, which, in turn, gets oxygen from the atmosphere. Even in patients without aniridia, a soft contact lens serves as a barrier to oxygen supply for the cornea. Over time, especially with extensive wear, this can result in abnormal blood vessels growing onto the surface of the eye. This situation is more problematic for the person with aniridia. It turns out that folks with aniridia are born with a reduced capacity to re-create the cells that form the surface of the cornea (so-called, limbal stem cell deficiency).

Over time, what can happen in people with aniridia is that abnormal cells that are not transparent can grow onto the cornea. This can be accompanied by new, abnormal blood vessel growth. A contact lens therefore makes a tough situation a little tougher. It reduces corneal oxygenation, making the blood vessel growth potentially worse. It also may cause little areas of "micro-trauma", which, over time, speed up the process of the corneal disease. Some patients with aniridia also have dry eye. This can make the contact lens wear both

uncomfortable and increase the "microtrauma."

If a contact lens is required for some reason, consult your eye care professional as to the best brand. It turns out that some brands of contact lenses allow more oxygen through than others. New things are constantly being introduced in the contact lens market, so it is best to work with your doctor in real time on choosing a brand.

**Q: So protecting the aniridic cornea is very important. What are some other ways to do this? (use of preservative-free eye-drops/lubricants? catching eye infections promptly and treating them with antibiotics that are gentle to the cornea?)**

A: Protecting from light can be easily done with eyeglasses. Many plastics will block at least some of the UV spectrum light (but not visible light) that is received by the eye and UV-rated sunglasses will do the job much better. Ideally, these lenses would at least partially "wrap around" to protect the eye from light coming from the side. No one has shown for sure that light damaged the eye of patients with aniridia. However, it seems reasonable to do simple things like sunglasses that might help and don't do any harm.

Using frequent artificial tears can help keep the cornea moist. In patients with diagnosed dry eye, sometimes things like occluding the tear drainage system (the puncta) can be helpful. Most cases of eye infections in adults are viral, not bacterial. Therefore, it is in your best interest to only take antibiotics prescribed by an eye care professional (not your general practitioner), as they may not be doing any good. In fact, some antibiotics have the side effect of making the surface of the eye abnormal, which could potentially exacerbate things in a patient with aniridia.

Probably the best advice is to be under the care of an eye care professional who is familiar with aniridia. You should be followed regularly.

**Q: Cataracts are extremely common in people with aniridia. When**

**is it appropriate to consider removal? Are there any factors specific to aniridia that should be considered (ie, balancing risk of exacerbating keratopathy, different surgical approaches, considering concurrent placement of artificial iris, etc)?**

A: It is appropriate to remove a cataract (an opacity in the lens of the eye), when it either reduces the patient's vision, is leading to significant problems with glare, or is so severe as to interfere with your doctor's view of the retina. Surgery is often very helpful to patients with loss of vision due to cataract. Nowadays, cataract surgery is less stressful to the eye than it once was. The incisions are smaller, the instruments and solutions used are better, and there are many options for intraocular lens implantation. However, it still is some stress to the eye, and should be done by a surgeon with a lot of experience. If a patient is interested in having an artificial iris implanted, he or she should go to a doctor who has a lot of experience with this surgery. I would also suggest getting more than one opinion. To my knowledge, there is no well-controlled trial showing that an artificial iris is overall better for patients with aniridia. While patients may experience some improvement with glare and cosmesis, there is also a risk of intraocular inflammation and bleeding after the surgery. These are additional "stressors" to the eye.

**Q: Glaucoma is a big problem for many people with aniridia. What are some of the issues in diagnosing and treating glaucoma associated with aniridia, and how might these problems be overcome? (diagnosis: implications of increased corneal thickness in aniridia...inaccuracy of pressure readings due to anesthesia [for EUA] Treatment: glaucoma surgery in aniridics...differences between latest generation Molteno vs. other implants?)**

A: The diagnosis of glaucoma in patients with aniridia can be tricky for a number of reasons. First, in the case of children, it is hard to get an accurate intraocular pressure in the office. We often use other "markers" for glaucoma to help us follow a child, including the diameter of their corneas, whether they are becoming



more nearsighted, and how the optic nerve looks on exam. We also do exams under anesthesia, in which we can measure eye pressure. However, the anesthesia itself can affect the eye pressure, so it is important for doctors to really look at whole picture, not just the number received from a pressure measurement. Second, folks with aniridia can have abnormal thicknesses to their cornea. While this does not really affect their day-to-day life or vision, it can affect the pressure measurement. In particular, thicker-than-average corneas falsely elevate the pressure measurement. The treatment of glaucoma needs to be tailored for the individual patient depending on his/her age and history, as well as by the severity of the disease.

**Q: The artificial iris and artificial cornea are some of the newest treatments for people with aniridia. Where does this research stand currently (risks/benefits)?**

A: Keratoprotheses (artificial corneas) have made significant advances in the past years. In general, these would only be considered in patients with fairly opaque corneas, in whom other surgeries had either previously failed or would not likely succeed. Again, whether to proceed with a keratoprosthesis is complicated and should be done on an individual basis with a surgeon who has a lot of experience with these devices. In general, the people who benefit the most from this option are those who have purely corneal disease. Because the vision of folks with aniridia is also impaired by foveal hypoplasia, nystagmus, and/or optic nerve problems, this should be taken into consideration when deciding on the timing and the likely benefit of surgery.

**Q: Until recently, advanced aniridic keratopathy was treated with cornea transplant. Unfortunately, long-term success with this treatment was low. Stem cell transplants for this condition are becoming more common, and seem to have a better success rate. Why is this, and at what point should a patient consider stem cell transplant?**

A: The cells on the very surface of the cornea are continually renewed. The stem cells that are constantly making a new corneal surface are at the

interface between the edge of the cornea and the white of the eye (the conjunctiva). This area is called the limbus and the stem cells doing the work are called limbal stem cells. It turns out that these limbal stem cells are not sufficient in folks with aniridia.

Over time, the eye surface can become clouded as abnormal cells start to migrate into the clearer parts of the cornea. Part of the reason why corneal transplants often fail in people with aniridia is that the transplant does not replace these deficient limbal stem cells. Therefore, to increase success, pioneers in the field, such as Dr. Edward Holland, have developed techniques to transplant both limbal stem cells and a new cornea to improve vision. Dr. Holland's success with this approach has been superior to that observed with corneal transplant alone.

**Q: Are you currently involved in research on aniridia? What is your involvement with the NIH/WAGR Syndrome Study? (Will you personally examine study participants who attend Phase II of the study (go to the NIH)? What type of examination can participants expect? What are your goals for the NIH/WAGR study?)**

A: We are always happy to see patients with aniridia at the NIH. We are currently wrapping up an observational study in collaboration with Dr. Holland on the correlation between someone's mutation in PAX6 and the severity of their corneal disease. I am also involved in a study with Dr. Joan Han looking at the WAGR syndrome.

I am interested in knowing what correlation there is between the degree of chromosomal deletion and the severity of their eye disease. I also want to look at what is happening with the retinas of adult patients with aniridia. PAX6 not only plays a role in eye development, but it is also expressed in some cells of the retina in adulthood. Do patients with aniridia have subtle, but real changes in their vision over time because of this? We really don't know.

The kind of examinations done at the NIH are similar to those you would receive anywhere. We would do a

complete eye exam and perhaps pursue some more advance, non-invasive tests. We may ask for a sample of blood for the research. The overall goal is to understand the vision of folks with aniridia and to therefore better serve them.



Our sincerest appreciation to the March of Dimes Michigan Chapter.

The IWSA was given a Community Award, which will offset some of the expense for printing and mailing the 2008 "WINGS" newsletters.

**IWSA Officers & Board Members:**

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Acting Vice President - Karen Rose**

**Secretary - Tammie Hefty  
Assistant Secretary - Julie Dell**

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**Member - Shari Krantz**

**Health Questions?  
Families & Physicians please Contact:**

**Kelly Trout, RN, BSN  
IWSA Health Consultant & Liaison to our Medical Advisory Board.**

[KellyTrout@sbcglobal.net](mailto:KellyTrout@sbcglobal.net)



### In Loving Memory of Linda Mae Foss

Former Virginia resident Linda Mae Foss, 53, died Friday, February 8, 2008, at her group home in Chisholm, Minnesota.

Linda was born with WAGR syndrome, and was the oldest known WAGR survivor in the world. She was born December 7, 1954, to Rodney and Dorothy Foss, and had lived at home and in Brainerd before her move to the Kingston Group Home where she enjoyed her second family for the last 8 years.

She thrived and blossomed at Kingston due to the wonderful care given her by the amazing staff at the home and the support staff and supervisors at Northern Habilitative Services.

Linda put everyone that she ever met into her "memory bank" and each person she got to know enriched her life. She taught us more about life than we taught her.

Linda was preceded in death by her father, Rodney. She is survived by her mother, Dorothy Foss, of Virginia, her sister, Luann (Dr. Byron) Rowell of Grand Rapids; her nieces, Kristin (David) Heebner of Minneapolis, Stephanie (Harlan) Keppler of Hermantown, and Lindsay Rowell (fiancé, Gary Bednar) of Blaine; her aunts, Helen Gunderson and Audrey Martinson of Virginia, many cousins, and her friends and family at Kingston.

A private memorial service was held. The IWSA wishes to thank all of Linda Mae Foss' friends and relatives that made monetary contributions in her name.

### TO ALL PARENTS by Edgar Guest

"I'll lend you for a little time a child of mine," (God) said,  
For you to love the while she lives and mourn for when she's dead.  
It may be six or seven years, or twenty-two or three,  
But will you, till I call her back, take care of her for me?

She'll bring her charms to gladden you, and shall her stay be brief,  
You'll have her lovely memories as solace for your grief.  
I cannot promise she will stay, since all from earth return,  
But there are lessons taught down there I want this child to learn.

I've looked the wide world over in my search for teachers true  
And from the throngs that crowd life's lanes I have selected you  
Now will you give her all your love, nor think the labor vain  
Nor hate me when I come to call to take her back again?

I fancied that I hear them say: "Dear Lord, Thy will be done"  
"For all the joy Thy child shall bring, the risk of grief we'll run,  
We'll shelter her with tenderness, we'll love her while we may,  
And for the happiness we've known forever grateful stay;

"But shall the angels call for her much sooner than we've planned,  
We'll brave the bitter grief that comes and try to understand."

### The Dark Side of the Moon

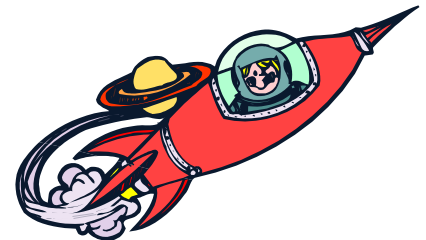
By Tammie Hefty

I love the movie Apollo 13. It's a great movie about facing the odds and coming out a victor. After a recent daily meditation I was reading, I started thinking about the similarities in dealing with WAGR syndrome and being on the dark side of the moon.

When we look at the moon from our earthly point of view, we see the side that is reflecting the sun's light. Since the moon is not just a flat disc, however, we know there is an equal side that is being obscured from the sun's rays, and that is the dark side of the moon.

In Apollo 13, the astronauts were trapped in the shuttle and did not have the energy they needed to fly the shuttle back to earth as they normally would. So one of the things they had to do was power down the shuttle to conserve energy, and let the gravitational pull of the moon move them further along. This required them to be on the dark side of the moon without any

warmth or light or contact with those at ground control.



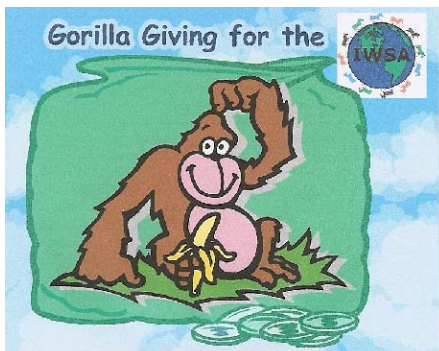
Our journey with WAGR syndrome can be a lot like that journey for the astronauts on Apollo 13. We started off the journey as parents-to-be, anxiously anticipating the arrival of our child, and visualizing what our lives were going to be like. But, something went wrong. In Apollo 13, something went wrong when one of the astronauts "stirred the tanks." In WAGR, something went wrong when the DNA was working along that mystical 11p (etc.) chromosome. Suddenly we find ourselves trying to gather some of the most brilliant doctors and/or rocket scientists together to figure out how to get us safely home.

We experience fear at times; a

distance, a loneliness that feels like the dark side of the moon. Our friends and family sit on earth, no contact with us, no radio, no TV, no pictures to tell them what we are doing, or HOW we are doing while we drift through space powered by nothing by gravity. In our own little shuttles, we huddle together and try to keep warm and try to keep the fear of the unknown buried away, because we know that the only way to combat the loneliness of the dark side of the moon is to hold tight to each other.

I'm so glad that I have my fellow astronauts of the IWSA, because we know what it's like being pioneers in space. We've all been on the dark side of the moon at one point or another. An ultra sound, EUA, or a seizure can send any of us right out of the sun's rays and around to the desolate darkness of the other side. Then we'll hear a story about a triumphant first day of kindergarten or see a picture of one of our boys in a tux, and the gravitational pull slingshots us a little closer to earth.

We are the pioneers of space, we have spanned the galaxy of experience and we know how to coach each other back to where we need to be. So, along with our titles of: gorilla mom or gorilla dad, boo-boo kisser, fear soother, song singer, laundry cleaner, linoleum scrubber, medicine dispenser, and appointment maker, we can add: space cowboy, been to the dark side of the moon and lived to tell about it.



Do you want to help the International WAGR Syndrome Association but don't know how?

## International WAGR Syndrome Association Profit & Loss January - December 2007

Totals	
<b>Income</b>	
Non-Profit Income - Corporate Contributions	\$ 284.70
Non-Profit Income - Other Non Profit	\$ 934.58
Non-Profit Income - Personal Contributions	\$ 6,062.41
Non-Profit Income - Retailer Contributions	136.47
<b>Total Non Profit Income</b>	<b>\$ 7,418.16</b>
<b>Total Income</b>	<b>\$ 7,418.16</b>
<b>Expenses</b>	
Conference Fees	\$ 75.00
Technology - website	\$ 438.95
Gifts for Sick Families	\$ 261.04
Legal & Professional Fees	\$ 225.00
Board Business	\$ 68.29
Miscellaneous	\$ 15.13
Mailing/Postage	\$ 872.60
Organizational Fees	\$ 25.00
Rent or Lease	\$ 60.00
Stationery & Printing	\$ 1,259.72
Newsletter	\$ 696.52
Supplies	\$ 102.37
Taxes & Licenses	\$ 95.00
Travel	\$ 137.65
WAGR Weekend Family Assistance Scholarship	\$ 1,982.80
WAGR Weekend	\$ 7,093.00
<b>Total Expenses</b>	<b>\$ 13,408.07</b>
<b>Net Operating Income</b>	<b>\$ 5,989.91</b>
<b>Net Income</b>	<b>\$ 5,989.91</b>

Well, the Gorilla Giving for the IWSA label was created for you. Do you have spare change that you don't know what to do with? Who likes having all that coin weighing you down? Simply paste this label on an empty jar or container and use it to collect all your loose change. At the end of a few months, simply count it up and donate it to the IWSA. This is a great way to get the entire family involved. We have included a label for you.

Just a reminder - there are several easy ways people can help raise funds for our organization.

We are able to accept donations through special designation with United Way. Our designation number for EWay participants is 822798, otherwise you must designate International WAGR Syndrome Association by name using the donor option program.

Looking for some IWSA logo items or apparel? Why not visit our Cafepress online store.  
<http://www.cafepress.com/wagr>  
 A portion of the proceeds from all sales will be donated to our organization.

Next time you need to order your special someone flowers try visiting <http://wagr.flowerpetal.com> Here you can purchase flowers and gifts for birthdays, graduations, anniversaries and holidays. You will feel good knowing that with every purchase you will be supporting our efforts.

The Little Tikes Giving Program makes it simple for you to shop online for Little Tikes children's products and earn a 5% cash reward for the International WAGR Syndrome Association. Simply click on the Little Tikes tab at [www.wagr.org](http://www.wagr.org) for more information.

Donating by credit card is easy to do too. You can do this by typing in the following link or by going to [justgive.org](http://justgive.org) and searching for IWSA.

<http://www.justgive.org/giving/donate.jsp?charityId=19385&>

Catherine Milian is our newly appointed Fundraising Coordinator. If you would like to be a member of her team please feel free to contact her at [cathymilian@yahoo.com](mailto:cathymilian@yahoo.com)

**Thank you for your support!!**

**Winning Attitudes, Great Rewards**



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, Kosovo, New Zealand, Peru, Philippines, Poland, Puerto Rico, Romania, Saudi Arabia, Scotland, South Africa, Tanzania, Turkey, and The United States of America

Con't from pg. 1 ~ Chair

If you love to shop online, you can help support the IWSA by purchasing items through iGive.com. Choose from over 680 of your favorite retail stores. A portion of all sales will be donated to our organization when you select the IWSA.

We are very happy to be hosting WAGR weekend this summer here in Michigan. We would like to extend an invitation to all our families to attend. Please join us for fun, fellowship and good times.

On a personal note.....Nicholas is doing great academically and medically. He's had a few more colds to battle this winter but nothing compared to years past.

Clem took Nicholas and my niece to see Sesame Street Live last month. They had great seats, third row ~ center stage. They danced, they sang, they really enjoyed the entire show.

Nicholas is currently taking swimming lessons at our local high school pool. Like most of our kids, Nicholas is like a fish when it comes to water. He is trying to learn strokes but prefers to swim under water. I'm just glad he can get exercise from doing something he likes so much.

Right before Christmas I went with Nicholas and his class to The Henry Ford Museum in Dearborn, Michigan. The museum highlights the history of inventions over the years. Of course it houses many cars, trucks, life-size locomotives. It also contains huge war planes,

Presidential limousines, the Rosa Parks bus, Oscar Mayer Wienermobile and much more.

The kids didn't really understand or appreciate all the history but did enjoy taking part in the hands on displays. If you are a history buff and attending WAGR Weekend, you will want to put this on your list to see.

Take care everyone,  
All the best,  
Annie Prusakiewicz



Here is Nicholas taking his friends for a ride in a Ford Model T.

### "C3" Day another HUGE Success in Riverview, MI

One Friday every month is designated Casual Clothes for a Cause Day (C3 Day) in the Riverview Community School District. Staff members pay at least \$5 to "dress down" or wear jeans on this day. Money raised is donated to pre-selected charities.

The generous staff in the Riverview Community School District raised close to \$800 dollars for the IWSA in December.

### Way to go "Pirates" Thank you again for your generosity & support!

Do you have a fundraising idea or opportunity? Maybe your work place would like to host their own C3 day for the IWSA? We would love to hear from you. Please e-mail your ideas to [cathymilian@yahoo.com](mailto:cathymilian@yahoo.com)

**Yes**, you CAN make a difference!



### 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**

**Winning Attitudes, Great Rewards**