



WINGS

WAGR Information, News, Gorilla Stories

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

www.wagr.org
Fall/Winter 2008

From the President's pen

Dear Families & Friends,

This year has been another wonderful year for our organization.

I sit in amazement at the number of families from around the globe that have joined us in the past 8 years.

It brings us great pleasure to know that you are not alone. Many of us spent years without a connection and that truly was the driving force to create this organization.

We are pleased to be able to bring you all the latest information and events through this newsletter. Our website is continually updated with the latest information as it becomes available. We also provide gifts to hospitalized children. The most precious event each year is our WAGR Weekend gathering.

All are very important activities and we love being able to do it.

Our organization is currently able to provide the above men-

tioned resources free of charge. This is possible through generous personal donations, fundraisers, sponsorship, and a Community Award 2008 from March of Dimes Michigan Chapter.

We are a unique group in many ways and it has hindered our ability to secure grant funding to assist us in general operations or activities.

Please consider spreading the word about our mission and encourage others to support us.

"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"

For more information please

visit our website or contact us directly reachingout@wagr.org.

Hope you all have a happy and healthy holiday season.

*Be well,
Catherine*

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The Fairy Queen Written by: Elly Chapple

Well it's a fairy tale all right, there are the moments that have you on the edge of your seat and those that make you cry but like all good stories there's a lovely ending...

I remember being 36 weeks pregnant and having been told that the baby was head down, I was measuring 36cm then we were set to go. A by chance encounter with our consultant regarding my husband James' blood clotting problem and all eyes suddenly fell on me. 'How many weeks are you?' he asked eyeing my stomach. I protectively put my hand on bump and explained that 3 days ago I had all checks and was fine. 'Oh no, you're looking way too small, get on the bed please.' Heart in mouth, palms sweating I did so and after a fairly uncomfortable examination he said 'Well you're measuring 32cm and the baby is breech' - pause, shock etc - so what next? 'A scan and this afternoon, we need to have a good look'.

The scan couldn't be done until the next day and with James working, I nervously went off with my Mum to be checked. After two scans to ensure what they were seeing was right they came back to me with 'the baby needs to come out now really, the baby is small and there is Oligohydramnios showing'. So reluctantly and after a lot of persuasion - silly me, I didn't want to choose their birthday! - I agreed to a c-section the following Tuesday.

That weekend was surreal, the excitement about meeting our baby but tinged with what really was the problem and would there be more set in.

Tuesday came round fast and the epidural was fine. James was so excited dressed up in his white suit; he looked like a space man. The anaesthetist was amazing, so calm and helpful. Soon the baby was out and I remember just



shouting what is it? 'A girl' came the reply and my heart jumped - I had been hoping so!! The next hour or so was strange, meeting your little life after 9 months and seeing this beautiful little person words can't describe the feeling. I looked at James and he at me, 'What shall we call her then?' we had chosen Ebony and Ella, but one look at this precious bundle and I knew she was a Fairy Queen - so Ella it was.

After about 8 hours she still wouldn't feed and so she was transferred to special care and I was given a spare room of my own, thankfully. I just couldn't bear the thought of listening to all the mums with their babies on the ward and mine was taken away.

The next week was a flurry of emotion and the start of a real rollercoaster ride. I saw her every minute I was allowed to and it was hard not being able to hold her all the time.

No one prepares you or tells you how scary becoming a Mum can be - yes it's amazing but it's terrifying too. What to do, how to do it and suddenly you have this huge responsibility. On the third day while James was visiting us, he noticed a white spot on her left eye - I think I was quite low at this point after the whole birth shock and I refused to think it was anything. Thank goodness he was so calm and insistent. Sure enough the ophthalmologist was called and suggested it could be cataracts so we would need to be seen in clinic fairly soon. Fear set

in that day and my lioness streak seemed to come right to the surface. Next was an appointment with the Consultant on call. She sat us down quite calmly and proceeded to say that they thought Ella was Downs Syndrome owing to her single palmer creases and that more tests would need to be done, primarily genetic to confirm any diagnosis. Again that fear feeling crept in and it took all of my might not to smack the woman as she sat on the floor and spoke patronisingly to James, referring to me as 'Mum' in the second person, like I wasn't there, I wasn't coping. I was in shock - what did she expect?

Our walk into the unknown started to take shape; tests abounded and sure enough finally they came back with the diagnosis 'She's got WAGR syndrome' err what? I don't think there was one doctor we saw who really knew anything about it. 'Yes it's rare and this is what you can expect.' Roll on the list of conditions...our rational selves took over, we could do this, we were strong, we were invincible and they were wrong.

At times like that children already seem to know when you need a lift and she promptly pulled the feed tube out of her nose and pushed the hat off her head - she was 3 days old, I was so proud, thanks baby I thought with a smile.

Things continued normally and eventually we were transferred to the midwifery suite nearer to home where I began to relax and I think she did too. For a very strange reason - why I so strongly believe in mother's instinct - I wouldn't open the curtains or have nay bright light around her. The nurses would come in and say 'This baby is never going to know what daylight is!' and throw them open. As soon as they left, I closed them again and turned the lights off.

Our appointment rounds started, the endless poking and prodding, the consultants, the explanations, life was a whirl. We found it very difficult at times to be happy like new parents should be - everyone

else seemed to be enjoying their newborns. I would take her to baby group and be terrified, sat there with my tiny tot who couldn't open her eyes or feed well and look at these healthy bouncing things that were already doing amazing thing. Oh, the baby Olympics - don't they drive you mad? Having to explain to people was awful until you get to grips with it. It took a long time. I slid into post-natal depression and took an overdose when it got so bad my head just couldn't take it anymore. I wasn't really trying to die, just desperate for someone, anyone to help me and say - this is damned hard and it's ok not to be ok.

It was the beginning of the start to being happy; sometimes you really do have to go all the way down to the bottom to come back up. Friends were amazing, family were so strong for us and we slowly climbed out of the hole. There were dark times and bright ones. Our learning curves were extreme to say the least. By 8 months old Ella was still the 'snuffly' baby and was weaning well. I decided to take her on holiday with my best friend and twins to Portugal as James was working abroad quite a lot. Company was good and that of a close friend was the best.

Our 10-day break was lovely but smattered with a 4-day hospital stay in Portugal where Ella was suctioned with tubes, given endless salbutamol and various antibiotics. At one stage she was taking 5 different medicines. She couldn't sleep, had an awful cough and was stopping breathing a lot.

Portugal is a lovely place but I didn't feel safe in the medical hands especially not speaking the language. Finding the WAGR network and the Mums, saved us so much - there finally, people had a handle on this thing and how to deal with it, it was ok to talk about anything and they would listen, the advice was invaluable and it gave us great confidence to do what we instinctively felt was right for Ella.

It was a good start to a journey that took us in and out of hospital for the next 4 months until we decided to go privately to Great

Ormond Street before our team here suggested a tracheotomy. Absolutely no way I said, her ability to be vocal, loud and sing could not be taken away, she would lose interest in life I was sure. She had so little movement available to her - her greatest pleasure was being very loud!

Great Ormond Street removed her adenoids, started her using a nasal pharyngeal airway and for the first time in a year our baby slept soundly. Her oxygen saturations must have been so low, she had a huge will to live and fought with all her might, they said we were very lucky to still have her with us.

Since that time she had grown, we've discovered her reflux, she's managing to eat semi-pureed food and she's now cruising around the house - at great speed!!! She's got constant ear infections and coughs etc but we turn up at our open access ward now and ask for the relevant antibiotic. It's comical really, we've become the best doctors she can have, knowing her inside out and being able to read the signs early enough.

She's coming up for two now and has brought an immense amount of love to both our families and indeed healed so many wounds between members. She truly was sent to us all for a reason and she is so special. There are those nights when you sit and think 'why us, we can't go on, we're exhausted, look at everyone else, it's so easy' but then you get the balancing days of 'wow, she's amazing and so interesting and brave and well - we're honoured that she chose us'

So there it is, very long-winded, fairly medical, but thanks for letting me write, it's been cathartic and healing to do so and face some things I'd kept buried. Life can only go up now and we truly are blessed to have our little Fairy Queen.



Gorillas Give Back

In the Spring Newsletter the idea of the Gorilla Giving Label was born. It was a way to give money to the IWSA and not even feel like you were doing anything. We immediately started a jar for our family to put our spare change in. It was simple....go to the store, have change from your purchase, throw the change in the jar. We found change in the car and even in the couch cushions. The kids had so much fun finding change to put in the jar. Little by little the amount increased, that's when we had an idea.

We emailed everyone we knew a letter with the Gorilla Giving Label attached. We asked them to participate with us in this simple fundraising event. Our plan or should we say "hope" was to take at least \$500 of donations from family and friends to WAGR Weekend 2008 in Michigan. Much to our surprise, our family and friends rallied to help us accomplish and surpass our goal.

We presented 18 checks totally \$1,860 in honor of our son Hayden to the IWSA. We were so thrilled and excited to give back to an organization that got us through



Forward newsletter submissions, stories, pictures or ideas to:

Annie Prusakiewicz
P.O. Box 392
Allen Park, MI 48101
E-mail: TheMooZoo@aol.com

our toughest days as new parents of a child with WAGR Syndrome. Our family would be lost without the IWSA. We are truly blessed to have found such a caring, helpful, and life saving organization.

Julie & Brian Dell

Special Thank You To Our Donors:

The Home Association of
McSherrystown
ESAB Welding and Cutting, R&D
Cedar Hill Archery
Steve & Monica Wright
Nikki & Neal Hoffman
Johnny & Lorrie Hyrcek
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Barbara & Francis Weaver
Shannon & Brian Cromer
Barbara & Ray Merritt
Rodney Bixler
Tania & Ken Kuhn
Tom & Mary Schuchart



Moving?

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

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

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

W-Wilms' tumor

A-Aniridia

G-Genital and/or urinary tract abnormalities

R-mental retardation/developmental disabilities

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

To My Darling, Megan

Written by: Vicky Jones

What can you see
little black-eyed one?
Can you focus in the dark?
Does it hurt in the sun?

You find a tiny smartie
That you've dropped on the floor
Then turn around so quickly
And go straight into the door.

"Sit in the corner" they said
But what do 'experts' know?
You shuffle around the furniture
Not understanding the meaning of slow.

And so you've struggled so long
to get this far.
Your determination to succeed
Has made you what you are.

Your cheeky little grin
tells me there's something up your sleeve.
You always try so hard
and in trying, you achieve.

You babble, pull and point
to get yourself heard.
That's why it's such a joy
when you manage another new word.

Special children for special people,
Isn't that what they say?
Well, I'm not all that great
But Meggie, you'll make it one day.

Each little occasion is a bonus
You have so much love to share.
What you give me is such a treasure
And that's what makes me care.

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.

The story from the University of Miami group



With the strong support of the IWSA families, we have completed our studies on 31 patients with WAGR syndrome in collaboration with Dr. Han at NIH. We have summarized our results and an article has been accepted for publishing in the journal of Cytogenetic and Genome Research. From the clinical data collected, it was noted that most of the patients have typical clinical features of WAGR and many also have behavioral problems including autism. The main purpose of this study was to find out what has caused mental deficit and autistic features in our WAGR kids.

We knew that the Wilms tumor and genitourinary anomalies are caused by missing a copy of the *WT1* gene and the eye problems are caused by missing the *PAX6* gene. However, we did not know what genes have contributed to the abnormal mental development and/or autism. We used a technology called microarray based comparative genomic hybridization to study the whole genome with a particular focus on the WAGR region on chromosome 11. With 40 thousands of DNA probes placed on each microarray, we characterized the chromosome 11 deletions in 31 patients and identified all the genes involved in each deletion.

We found that the size of the deletions was different in each patient, ranging from 4.9 to 23 million base pairs. The number of genes that were missing varied from 18 to 62 with an average of 40 genes. In



addition to the *WT1* and *PAX6* genes, we noted that all the patients studied lost a copy of a gene called *PRRG4* which stands for “transmembrane gamma-

Dr. Fan with kids, WAGR Weekend 2005

carboxyglutamic acid protein 4”.

The majority of them also lost a copy of a gene called *BDNF*, a “brain-derived neurotrophic factor” and a copy of another gene, *SLCIA2*, the “solute carrier family 1 glial high affinity glutamate transporter member 2”. We found that missing a copy of the *BDNF* and the *SLCIA2* gene occurred in patients with autism more often than in those without autism. Because these genes play important roles in normal development and cognitive functions of the brain, our studies have suggested that missing a copy of the *SLCIA2*, *PRRG4*, and *BDNF* genes can be the cause of mental retardation and behavioral problems in WAGR patients.

In particular, we have proposed that the *BDNF* gene may modulate the risk of autism in WAGR patients because this gene is regulated by a gene called *MECP2*. This simply makes sense as mutations of the *MECP2* gene causes Rett syndrome which is also associated with autism. I would like to say thank you so much, to all the families who have contributed to this study.

MICHIGAN MEMORIES



The highlight of WAGR Weekend for me each year is seeing our children together. There is this incredible chemistry when the children meet each other as well as the siblings. It is a silent understanding they all share, just as we do.

Meeting new families & catching up with old friends is a big thrill to say the least. I learn so much from each family and their personal experiences. Talking over the internet does not compare to spending time together.

On the plane ride home I began counting how many children I have met with WAGR syndrome. I have had the pleasure of meeting 34 children.

I can remember being told that I would never meet another child like Irm.

Who knew?

Catherine Luis, N



Irma & Catherine

Here are a few of my things I will remember...

Danica....I must have gotten 100 kisses from that sweet princess.



Nicholas & Hollywood Hayden

Grace Farren



Johanna Meyer

Kelsie Brooks

Danica



Jenna & Abby

She is such a cuddle-bug. Danica and Hayden held hands during the kid photo...too cute!

Grace....This girl has all the dance moves. I was very impressed at her talent of drinking water with about six straws at once!

Jenna....Always has a camera in hand and wants to take every-ones picture. Jenna-girl you are always a delight to see.

Nick....This guy is too funny. Hayden and Nick had fun the first night running around the room and I have some good pics of them dancing.

BTW the food Sunday was delicious Chef Nick. Let us know when your restaurant opens!

Kelsie...Such a delightful young lady. Everytime I would see her, she would always be smiling. She is so very polite too! I cannot believe she is sixteen already. This girl is going to do great things.

Johanna....Looked amazing. Her hair was so long compared to last year. You would never believe by looking at her everything she has been through. This little angel is such a miracle!



Nathan...Didn't really get to see Nathan too much :- (But he looked great. Of course Miss Irma was taking good care of him!

The Irm....Her memory of everyone's name amazes me. She is such the social butterfly and a hoot to be around.

Julie Dell, PA

I always look forward to WAGR Weekend each year. There is something about getting together with other families where you don't have to explain your child (or yourself for that matter) that provides great encouragement. This year we drove from our home in the Philadelphia area to the Detroit area. What fun to arrive at the hotel and see Nicholas Prusakiewicz ready to get the party started!

This year my parents were able to attend. They have always been supportive of our family and Jenna's special needs. To have them meet other families with a WAGR child opened their eyes to a whole other side of living with WAGR. They are very excited to continue to learn how they can help their grandchild, as well as attend another WAGR Weekend.

Jenna was thrilled to have her eleven year old cousin Abby attend the weekend with her. Abby has already asked if she can



GORILLA MOMS



GORILLA DADS

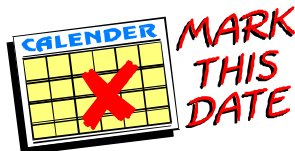
attend WAGR Weekend next year, because she made so many friends and feels like she understands her cousin better.

What happened at the weekend to cause this response in my extended family? It was each of the families in attendance. Each family, in their own way, demonstrates love and acceptance and perseverance and compassion and determination and hope.

Those qualities are fleshed out as each family eats a meal or swims or sits in a meeting. When someone has a need, another family jumps in to help. One WAGR child applauds another when hearing of an accomplishment. A father puts his arm around another father to let him know that he is not alone. A mother smiles at another mother to share in their joy. When someone needs to talk, there are several empathetic ears to listen.

Will we attend WAGR Weekend 2009? You better believe it! It is the jolt of refreshment that keeps me moving forward. The anticipation of seeing our friends who understand our life is something I treasure all year long.

Kelli Cox, PA



**THANK YOU!
WAGR WEEKEND 2008
DONORS**

- The Henry Ford
- Sam's Club (Southgate, MI)
- Outback Steakhouse
- Gary Printing
- False Alarm
- Classic Auto Body, Inc.
- Ames, DJ Service
- Ramada Inn
- Build a Bear
- Taylor Lanes
- Volunteers: Vana K., Rachel M., Audrey D., and Brandon D.

**WAGR Weekend 2009
Gaithersburg, Maryland
July 10-12, 2009**

SpringHill Suites by Marriott
Event Name: WAGR
WEEKEND
(301) 987-0900 or (888) 287-9400

Meetings & activities will be held at Bohrer Park Activity Center & Park

Tentative Itinerary:

- Friday Night** - Meet & Greet, Mom's night out
- Saturday** - NIH/NICHD Research Update from Dr. Han & Kristen
- Lunch - Catered
- Dinner - Catered pizza party, Dad's night out
- Free time - Playground, Water park, Miniature Golf, Skate Park
- Sunday** - Morning tour of the NIH
- Details and RSVP at the website <http://www.wagr.org/wagrweekends.html>

Dear WAGR Families,

We have many exciting updates to share about WAGR syndrome research here at the National Institutes of Health in Bethesda, Maryland.

First, we would like to introduce ourselves to anyone who is new to our research. 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. Kristen Danley, BS, is a research assistant working with Dr. Han as the coordinator of the WAGR syndrome study. She graduated from Bucknell University in central Pennsylvania with a degree in biology.

The current research we are conducting at the National Institutes of Health consists of a Phase I (ongoing) and a Phase II study (currently launching) that are designed to study how genotypes (the specific genes deleted) are associated with phenotypes (the clinical symptoms present) for each individual with WAGR. For both Phase I and Phase II of the study, we request 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.

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. WAGR patients of any age may enroll in Phase I. So far, the Phase I study has 54 families enrolled, and we would like to share some of the results that have come from the Phase I study so far.

Two abstracts on WAGR were presented at scientific meetings this past year. The first, presented at the Pediatric Academic Societies meeting in May, was entitled "BDNF Haploinsufficiency in Patients with WAGR Syndrome is Associated with Decreased Behavioral Responses to Pain." We reported that among people with WAGR syndrome, those who had deletion of one of the two copies of the gene for brain-derived neurotrophic factor (BDNF) appeared to be less sensitive to painful situations, like injuries and medical procedures, compared with those who did not have deletion of BDNF. The abstract can be viewed online at http://www.abstracts2view.com/pas/view.php?nu=PAS08L1_353.

The second abstract was presented at the Endocrine Society meeting in June and was entitled "Childhood Stature in WAGR Syndrome." We reported that people with WAGR syndrome tend to have normal length at birth but then develop shorter stature by the time they are toddlers and progress to have even shorter stature when they are teenagers, such that their final height as adults is approximately 1.5 standard deviations lower than the national average height for healthy adult men and women. The abstract can be viewed online at http://www.abstracts2view.com/endo/view.php?nu=ENDO08L_P2-576.

Finally, an article entitled "Brain-Derived Neurotrophic Factor and Obesity in the WAGR Syndrome"

was published in the August 28, 2008 issue of the New England Journal of Medicine. We reported that among people with WAGR syndrome, those who had deletion of one of the two copies of the BDNF gene had a predisposition to develop increased appetite and obesity compared with those who did not have deletion of BDNF. A summary of the article can be viewed online at <http://content.nejm.org/cgi/content/abstract/359/9/918>

The Phase II study is currently being launched, and we expect to be able to schedule the earliest visits for early November 2008. The Phase II study is entitled "WAGR Syndrome and Other 11p Contiguous Gene Deletions: Clinical Characterization and Correlation with Genotype," and children older than 6 years and adults with WAGR syndrome can participate. The study involves a one-week visit (arriving Sunday evening and leaving Saturday) to the NIH in Bethesda, MD during which participants stay on the NIH campus.

During the week, participants will receive a comprehensive evaluation of a variety of symptoms associated with WAGR syndrome. The evaluation includes testing of how much energy they use; testing of how they process glucose; evaluation of hearing; evaluation of vision; evaluation of behavior and motor skills; kidney screening; head and body imaging; dental and facial measurements; testing of sensation thresholds; and measurements of body composition (fat mass and non-fat mass). When we begin enrolling participants, we will include a full listing and description of the tests and procedures that will be done.

If you are interested in participating in the Phase I and/or Phase II studies or have other questions about research at the NIH, please feel free to contact Kristen or Joan and we would be happy to talk to you!

Thank you very much!

Kristen Danley: (301) 402-6762; danleykr@mail.nih.gov
Joan Han: (301) 435-7820; hanjo@mail.nih.gov

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Q. Why is the Phase II study open only to adults, teens, and children older than 6 years?

A. The study includes several tests that require blood draws. The National Institutes of Health has guidelines limiting the amount of blood that can be drawn for research purposes in children. In order for us to do our tests, children need to be at least 6 years old.

Q. How long is the Phase II study expected to go on?

A. The Phase II study is expected to be accepting participants for approximately three years or longer (through fall 2011 and possibly beyond). We also expect that further studies may arise as a result of the Phase II study, so we will continue to update families about future opportunities.

Q. If my child turns 6 during that time, can we join the Phase II study then?

A. Yes. Once your child is six, he or she may participate in the study. As your child's birthday approaches, feel free to contact us to begin discussing study participation and visit dates.

Q. My child is older than 6, but he is not verbal/has very limited speech. Can he participate in the Phase II study?

A. A child will not be disqualified from the study because of limited speech, but we will discuss participation with each family to ensure that the child will be able to participate in enough components of the study to make the visit to the NIH worthwhile for the family. Alternately, some families may prefer to wait until their child is a little older to participate. If you have any concerns or questions about your child's ability to participate, please contact us at any time.

Q. Is the Phase I study still open to enrollment?

A. Yes, the Phase I study is still accepting new participants. To participate in the Phase I study, a family completes a blood draw for biochemical and genetic testing (the blood draw can be done at a family's home or physician's office) and sends medical records to the NIH. If you are interested in enrolling or have questions about participation, please contact us for enrollment materials. There is no cost to participating in either the Phase I or Phase II study.

Q. We are already enrolled in the Phase I study. Do we have to enroll in Phase II separately?

A. The Phase II study does have separate enrollment, but the amount of paperwork you need to fill out is minimal if you have already completed paperwork from the Phase I study.

Q. Would the NIH like to continue to receive information from families about their children after completing the blood draw for Phase I or the visit for Phase II?

A. Yes! We are very grateful to every family participating in either or both of our studies, and we hope to maintain a long-term relationship with each family. We appreciate receiving medical updates (new diagnoses, medications, and developmental milestones reached) as well as social updates about graduations, achievements, travels, and any other news. We especially enjoy receiving photos, and we thank all the families who have shared such special moments with us.

Q. Why should my child participate in a research study? What are the benefits?

A. The purpose of our research is to learn more about how the

deletion of specific genes in people with WAGR syndrome results in specific clinical symptoms. By learning more about the function of the genes on chromosome 11, our hope is that our research will lead to improved health care for people with WAGR syndrome. Our goal is to permit medical care to be custom-tailored based on an individual patient's deletion profile. Your child's participation in this project may result in no direct benefit to your child, but will contribute to our knowledge about WAGR syndrome and other 11p deletion syndromes. Also, you will receive all the results of the clinical evaluations for your child.

Q. What are the components of Phase I versus Phase II of the study?

Phase I

- open to anyone with WAGR syndrome
- all ages
- may live anywhere in the world
- involves sending blood sample and medical records to the NIH
- participants receive results of all clinical laboratory and genetic testing
- no cost
- ongoing since 2006

Phase II

- open to anyone with WAGR syndrome *or aniridia*
- must be 6 yrs of age or older
- involves a one-week trip to the NIH in Bethesda, Maryland, USA (near Washington, D.C.)
- includes a wide variety of medical evaluations
- participants receive results of all clinical tests and evaluations
- no cost
- participants from outside the USA must provide

- own transportation to USA
- travel from within the USA to the NIH is covered
- lodging is provided
- compensation for time provided
- begins November 2008

Where Are the Old Ones? Life expectancy in WAGR Syndrome

by: Kelly Trout, RN, BSN
IWSA Health Consultant

Life expectancy in people with WAGR syndrome has not been studied, so the only information currently available comes from the members of this organization, which includes approximately 150 individuals with WAGR syndrome. The great majority of these are children, and the oldest living person we're aware of with WAGR syndrome is currently in her 40s.

There are several possible reasons for what appears to be a lack of older adults with WAGR syndrome.

The first possible reason is that effective treatment for Wilms tumor did not exist until the late 1970s. Children with WAGR syndrome have a 50% chance of developing Wilms tumor, so

of those born before 1980, only those who did not have Wilms tumor would have survived past early childhood.

A second reason is that "WAGR syndrome" has been known by many names over the years. Before the mid 1980s, it was known only as sporadic aniridia. But the availability and accuracy of genetic testing has increased over the last two decades, and various other terms have been used. These include: "Aniridia Wilms tumor syndrome," "AGR syndrome," "11p-syndrome," and "11p13 deletion syndrome." The designation, "WAGR syndrome" has only been in widespread use for about the past 10 years. For this reason, it is very likely that there are adults with WAGR syndrome whose diagnosis is listed instead as one of these older terms.

Another factor to consider is the age of the parents of adults with WAGR syndrome. People who are in their 60's or older are far less likely than younger parents to use a computer to look for information about their child's disorder, and so may be unaware of this organization. In addition, older age groups are also much less likely to join a support group, possibly because such groups did not exist when their children were young, or first diagnosed.

Finally, medical diagnosis and treatment of the conditions associated with WAGR syndrome has improved dramatically in the past twenty years.

These improvements include

monitoring for Wilms tumor every 3 months starting at birth, and testing for Focal Segmental Glomerulosclerosis (FSGS, a kidney disease associated with WAGR syndrome). This kidney disease occurs in up to 60% of individuals with WAGR syndrome, and can lead to kidney failure. But early diagnosis and treatment may significantly prolong the life of the kidney(s). In addition, dialysis and kidney transplants have proven successful in extending the lives of people with WAGR syndrome and FSGS.

While people with WAGR syndrome are at risk of early death from some of the medical complications of this disorder, progress in knowledge about these complications continues to improve. Today's children and young adults with WAGR syndrome are already benefiting from this progress, and their futures will be brighter and longer as a result.

To learn more about appropriate health care for people with WAGR syndrome, look here: <http://wagr.org/physicians-guide.html> A Parent's Guide to medical care is also available at the www.wagr.org website.



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.

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

We had representatives from 10 families at WAGR Weekend Michigan. Friday night all of our families gathered for an informal Meet and Greet sponsored by Frank Farren & False Alarm.

We enjoyed seeing all of the familiar faces and had a great time meeting the new families.

The Franceschini family and Aunt Heather traveled 15 hours from Northern Ontario Canada to attend WW. Little Danica was a gem. Everyone instantly fell in love with her. She was not stingy either when it came to doling out hugs and kisses. Of course, we all wanted to take her home.

The annual Moms W(h)ine and cheese night gave the moms time to get together and talk. We had even one Grandma, Karen (Jenna's Grandma) join us. By then end of the night Aunt Heather was ready to break into song, by Cher. We all had a great time and lots of laughs sharing stories of our children.

Saturday morning Helen Lane, with her years of experience in raising a child with WAGR and also her years with the Tennessee Early Intervention system spoke to our parents on the importance of Wills/Trusts and navigating the IEP system. The information was well received by all our families. The All U Can Eat Deli lunch gave our families the fuel to get ready for the Bowling family fun activity.

Many of our kids/parents joined us at Taylor Lanes, our local bowling alley to burn off some energy with a couple games bowling. We all had a great time. You should have seen Jim Meyer bowl holding baby Izzy. It was a remarkable sight. I think with the baby handicap, he took the top score of the afternoon.

Saturday evening we feasted on Pizza, garlic rolls and salad. We were all treated to the sounds of music, by Julie Ames (she is a full time high school teacher, but does DJ work on the side). The families enjoyed dancing to all the favorites - YMCA, Hokey Pokey, and Crocodile Joe. It was a great time had by all the young and dare I say "mature audience".

The moms took the kids so the dads

could gather for their annual Dad's Night out. Of course they complained about the interruptions, saying us women were trying to eavesdrop. I know the dads had a great time bonding and talking with each other.

Sunday morning was scheduled as free time. A few families left early so we never had the opportunity to say good bye. But they were missed. We gathered the rest of the families together one last time for a farewell dinner. Nicholas was the honorary chef of the afternoon and helped Chef Sam prepare the food for our meal. Chef Sam let him wear his hat and apron. Nicholas had a wonderful time. He had several assistants or Sous Chefs like Jenna Cox and Hayden Dell. We ate, socialized and then said our good byes. This is always the hardest part to the entire weekend. It's hard to say good bye to friends that you consider part of your family. Especially when they live so far away.

Many hours go into the planning an event like WAGR Weekend. I never really understood this until we chose to host the event for ourselves. Of course, none of this would have been possible if I didn't have the support of the IWSA board and its officers. A huge thank you to my wonderful husband and my two children. They helped me out tremendously or as much as I would let them. They were always there to lend a helping hand or offer suggestions. Special shouts out to Timothy at the Ramada Inn, his front desk staff, his hostess staff (Nicole & Kim especially), Chef Sam, Moe and Marvin for ensuring that all our families had a wonderful time. Chef Sam enjoyed our children and let Nicholas see what it was like to work in the kitchen. He honored special food requests like adding mushrooms to Jenna Cox's pizza and letting Nicholas give her whip cream for her apple pie. Chef Sam was genuinely concerned about our families and went the extra mile to make sure that all our needs were met.

I would like to thank Julie Ames for donating her DJ services so that our families could enjoy some music during our Pizza Party. It was nice to burn off some pizza calories by doing a little twisting and shouting. Build a Bear came through with a donation of some bears and I know our kids enjoyed getting their new play pals. To all the families, the grandparents and extended family that attended WAGR Weekend 2008, it would have not been a successful weekend without you. THANK YOU so much for just being here. See you all next year!!

Annie Prusakiewicz



Our Sibs: (Top) Casey, Maddie & Ashley Deegan, Casey & Bryce, Bottom Left - Hayden
Center - Danica & Catherine Bottom Right - Rockie & Connie Brooks

Winning Attitudes, Great Rewards

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

**Introducing our
IWSA Educational
Consultant**

Rhonda Sena is a co-founding member and former officer of our organization. She attended the University of New Mexico, receiving her Bachelor of Science in Elementary Education in 1993 and her Master of Arts in Elementary Education in 1994. She currently holds a K-8 teaching licensure, level 2 in New Mexico where she resides with her husband Alfred and their three children.

Rhonda acts as Educational Consultant to our organization, volunteers as a parent advocate, and just completed a grant with the US Department of Education that helped research and implement positive behavioral interventions for children with maladaptive social behaviors. Her son, Alex, is twelve and has WAGR Syndrome.

If you have any education related questions, Rhonda can be contacted offlist at CasaSena5@yahoo.com.

University of Miami Study

If you participated in the University of Miami study of ADD/ADHD and autism in WAGR syndrome, and are interested in receiving the genetic testing results please contact Ana Morales:

Phone: 1-305-243-9971 or Email: Amorales4@med.miami.edu

According to Ms. Morales, an article with findings from the University of Miami study has been accepted for publication, and she will let us know when it is published.



The Sena Family

**Big News: The MedQuest
Goes ONLINE!**

Some of the members of this group may remember "MedQuest," a questionnaire we mailed out in 2002 to help answer questions like "How many people with WAGR syndrome have (name a condition)?" The information we collected answered a whole lot more than that. It revealed

for the first time that WAGR syndrome often involves more than just W, A, G, and R. It also helped us build a set of guidelines for medical care that have now become the standard. In other words, we discovered that a small group of determined parents can change the world for their children.

Now it's time to do it again. Since that first MedQuest, our kids have grown, the group has grown a lot, and technology has grown like crazy. So this time, we're doing the MedQuest just one section at a time, and with a confidential, fast and easy ***online*** format.

The first section of the new MedQuest is called "WAGR syndrome: The Kidney and Urinary Tract" and it is now open. We hope that everyone will take a few moments and fill out this survey. To get started, just go to this link:

<http://www.surveymethods.com/EndUser.aspx?F2D6BAA2FAB6A4A5>



International WAGR Syndrome Association

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