International WAGR Syndrome Association

Annual Report 2011

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

WWW.WAGR.ORG

The Mission of the International WAGR Syndrome Association is to promote international knowledge and awareness of WAGR/11p Deletion Syndrome and its complications and treatments, to stimulate research, and to reach out to those affected by WAGR/11p Deletion Syndrome in an effort to improve their lives.

The Board members. Officers, and Leaders of the IWSA are

an all-volunteer group

dedicated to supporting and furthering the organization's mission.
In addition to having a loved one with WAGR/11p Deletion Syndrome,
each leader contributes skills, experience, and passion
to provide information and support to patients, parents, and physicians.

2011 Board members, Officers, and Leaders
Back Row, L-R: Leslie Volk, Annie Prusakiewicz, Kelly Trout, Rhonda Sena, Shari Krantz
Front Row, L-R: Julie Dell, Catherine Luis, Tammie Hefty, Jeff Hefty
Not pictured: Nikki Hoffman, Elizabeth Duffy, Vanessa Richter

2011 Financial Statement

International WAGR Syndrome Association
Profit & Loss
January - December 2011

	$
Income	
Non Profit Income	
Non-Profit Income - Grants	1,200.00
Non-Profit Income - Contributions	14,344.67
Total Non Profit Income	15,544.67
Interest Income	6.00
Total Income	15,550.67
Expenses	
Board Meeting	579.64
Computer Supplies	1,037.11
Conference Fees	450.00
Dues & Subscriptions	31.00
Gifts	205.00
Legal & Professional Fees	350.00
Mailing/Postage Business	966.57
Newsletter Expense	1,648.00
Office Expenses	351.33
Promotional - Constant Contact	210.00
Rent or Lease	40.00
Stationery & Printing	576.00
Supplies	189.16
Technology - Acct. Software	211.50
Technology - Website	174.49
Travel - Conference	440.95
WAGR Weekend Family Assistance Scholarship	1,275.85
WAGR Weekend Meals	2,171.69
WAGR Weekend Other Expenses	1,593.44
Total Expenses	12,501.73

In 2011…

Noah (OH) and Wesley (WA)

One of the most significant actions taken by the IWSA in 2011 was to announce modification of the name of the syndrome
from WAGR Syndrome to
"WAGR/11p Deletion Syndrome."

As medical technology advances, identifying specific genetic abnormalities is helping to further understand the causes of diseases and syndromes, including WAGR syndrome. According to Joan Han, MD, lead researcher for the WAGR study at the National Institutes of Health, "WAGR syndrome is classically thought of as having PAX6 and WT1 gene deletions, but "11p Deletion Syndrome" is a more encompassing diagnosis because it includes individuals with PAX6 and/or WT1 genes deleted, as well as those for whom other chromosome 11 genes are deleted." This name modification to WAGR/11p Deletion Syndrome helps to better clarify the genetic nature of WAGR Syndrome. In time, the disorder will become better known as "11p Deletion Syndrome."

Board Members, Officers and Leaders were privileged to pursue the mission and goals of the IWSA

IWSA Health Consultant Kelly Trout, RN, BSN continues to provide information to parents, patients and physicians on the medical conditions associated with WAGR/11p Deletion Syndrome. This information is shared via email, the IWSA listserv, telephone, newsletter articles, and the wagr.org website. Kelly also works in a liaison role with other patient advocacy and rare disease support organizations, including the Aniridia Foundation International, United Families of Aniridia, and the Aniridia Network UK, providing for example, a presentation on WAGR/11p Deletion Syndrome for the Aniridia Foundation's 2011 Make a Miracle Conference held in Atlanta, Georgia.

In 2011, Kelly attended the **"Genetic Alliance 25th Anniversary Annual Conference: 25 Years of Innovation"** participating in the Rare Disease track, as well as the Registry and Biobank Boot Camp. Attending this "boot camp" was the first step toward developing a **Patient Registry for WAGR/11p Deletion Syndrome.** Look for more information about this Registry in 2012.

Kelly also was recognized as an **Innovator in the Advocacy Community** at the Genetic Alliance 25th Anniversary Celebration held last September in Washington, DC. Additional activities included updating the "WAGR Syndrome" fact sheet for the **National Human Genome Research Institute;** updating "WAGR/11p Deletion Syndrome" for the **Rare Diseases Information Database;** and reviewing the "Clinical utility gene card for WAGR Syndrome," Clericuzio, C., Hingorani, M., Crolla, JA, van Heyningen, V, A, ***European Journal of Human Genetics,*** published online in January 2011.

IWSA Education Consultant Rhonda Sena, **M.Ed.,** continues to provide information and support to families and educational professionals on special education and positive behavioral interventions for individuals with WAGR/11p Deletion. Rhonda also developed and manages the **IWSA Lending Library**, which contains resources and books available for loan.

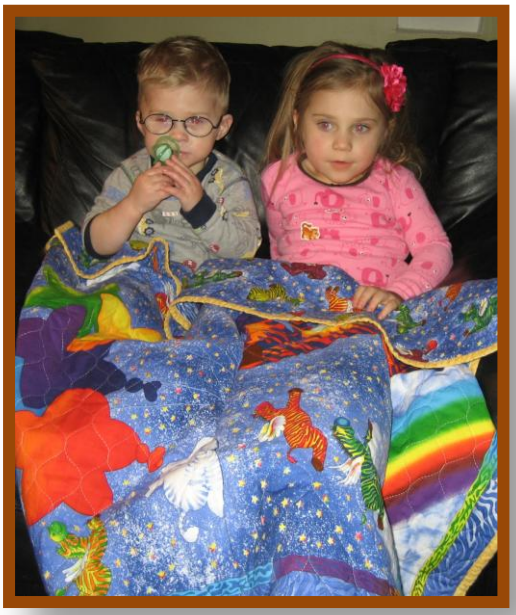
IWSA Board Member Shari Krantz attended the "**Genetic Alliance 25th Anniversary Annual Conference: 25 Years of Innovation**" Leadership Symposium in September. Shari also participated in the **Genetic Alliance/University of Washington** study on parent/patient advocate's experiences with genetic diagnosis. Shari represented the IWSA at the **National Institutes of Health's Celebration of Rare Disease Day 2011** in Bethesda, Maryland, which featured a presentation by Dr. Joan Han about the ongoing WAGR /11p Deletion Syndrome studies at NIH.

In addition to her duties as **IWSA Chairperson, Annie Prusakiewicz** also continues to edit the IWSA quarterly newsletter, "**WINGS: WAGR Information, News, and Gorilla Stories**," as well as the IWSA monthly online e-newsletter, "**Constant Contact**." Annie and her family also hosted the **2011 Annual WAGR Weekend** near Detroit, Michigan. WAGR Weekend is a vital part of fulfilling our mission to reach out to individuals and families affected by WAGR/11p Deletion Syndrome. It is a priceless opportunity to meet and spend time with other families who understand the challenges and joys of caring for someone with WAGR/11p Deletion Syndrome. **Thanks to many generous donors,** the IWSA was able to provide financial assistance to several families, enabling them to attend WAGR Weekend.

IWSA President Catherine Luis created and continues to manage one of IWSA's most important tools: **WAGR.org**. This award-winning website provides crucial, continually updated information and resources to patients, families, physicians, and educators.

In 2011, **Jeff Hefty moved from IWSA Treasurer to Board Member,** and will continue to help direct IWSA's mission and organizational direction.

In 2011, IWSA initiated a **Fundraising Committee, led by Vanessa Richter and Leslie Volk.** Vanessa and Leslie are coordinating efforts to fund the mission and activities of the organization.



Noah and Emily (OH) enjoy one of the ***IWSA Caring Quilts***

IWSA Vice President Tammie Hefty initiated and manages the "**IWSA Caring Quilts."** These beautiful zebra-themed quilts are shared in turns by families around the world. They are enjoyed for a time at home, in the hospital, or during outpatient procedures, then are washed and sent to another family. One quilt is kept at the NIH in Maryland, USA for individuals to use while participating in the WAGR/11p Deletion Syndrome Study. The IWSA Caring Quilts help families to know that they are not alone in managing the complexities of WAGR/11p Deletion Syndrome.

IWSA Secretary Julie Dell continues to welcome new families by providing "**Welcome Packets**" of information about the services and supports IWSA offers. Julie also manages the IWSA mailing and contact information lists, as well as IWSA correspondence.

IWSA Gift Coordinator Elizabeth Duffy continues to organize the cards and gifts sent to patients undergoing hospitalization.

Treasurer Nikki Hoffman provides management and reporting of **IWSA finances and budget**.

Throughout 2011, IWSA board members, officers, and leaders participated in a **wide variety of webinars (online workshops)** to increase their knowledge of information helpful to families and professionals.

Andy Choi (bottom row, center) and his family with the WAGR study team at the NIH. Clockwise from top left: Amanda Huey, Kim Choi, Tanvee Singh, Emily Yin, Matt Tsang, Albert Choi, Andy Choi, Kathy Choi, and Joan Han.

Research

Supporting and promoting research is a top priority for the IWSA. In 2011 we continued our partnership with the National Institutes of Health WAGR/11p Deletion studies. The initial study, under the direction of Dr Joan Han, began in 2006 with Phase I and in 2008 the clinical evaluation portion was opened. Several dozen families have participated in each phase of the genotype-phenotype study. The goal of the study is to learn how each patient's particular deletion affects their clinical symptoms.

In 2011, the WAGR study research team presented three abstracts at scientific meetings. The first, "**Duane Syndrome In WAGR Patients With CGH-defined Deletions Of 11p And Neuroradiologic Absence Of The Abducens Nerve**," was presented at the Association for Research in Vision and Ophthalmology Annual Meeting in Fort Lauderdale, Florida in May 2011. This abstract reports unilateral Duane syndrome (inability of one or both eyes to gaze outward) in three patients with WAGR syndrome, all of whom share a deletion in the 11p13 region. This suggests that a gene in the WAGR region may be important for the development of Duane syndrome.

The second abstract, "**Agenesis of the Olfactory (CN I) and Abducens (CN VI) Nerves in WAGR Syndrome**," was presented at the American Society of Neuroradiology 49th Annual Meeting in Seattle, Washington in June 2011. This abstract reports that absence or incomplete development of nerves in the brain, specifically nerves associated with smell (CN I) and outward eye gaze (CN VI), is not uncommon in patients with WAGR syndrome. This suggests that the genes on chromosome 11 around the WAGR region are involved in the development of these nerves in the brain.

Finally, "Morphologic **Alterations in Brain Structure in Patients with WAGR/11p deletion Syndrome**," was presented at the Organization for Human Brain Mapping's 17th Annual Meeting in Quebec City, Canada, also in June 2011. This abstract summarizes structural brain differences between patients with WAGR syndrome and patients with aniridia. The findings suggest that other genes on chromosome 11 besides *PAX6*, the gene that causes aniridia, may contribute to brain structure.

We are very excited to report that the research team recently received an NIH Bench-to-Bedside grant for a new clinical trial to study the effects of a medication that may replace some of the functions of brain-derived neurotrophic factor (BDNF) in patients who have deletions involving the gene that produces BDNF. Preliminary plans are to launch this new trial sometime in 2012. We are very excited about this new segment of the WAGR/11p-related studies and the IWSA plans to encourage and support IWSA families interested in participating.

The research being conducted at the NIH is providing vital information to guide treatment and improve the lives and health of individuals with WAGR/11p Deletion Syndrome.

Fundraising

Steve Wright
Winner of the 2011 50/50 Raffle

A major goal for the IWSA in 2011 was to create a fundraising committee. With IWSA families living across the US and in more than 20 countries around the world, we recognized the need to combine the interests and talents of many individuals. With the support and guidance of IWSA Board Chair Annie Prusakiewicz, the IWSA Fundraising Committee was formed with Vanessa Richter as its Chair. Vanessa is the aunt of a young child with WAGR/11p Deletion Syndrome, and has significant fundraising and leadership experience. Under Vanessa's direction, the Fundraising Committee met via conference calls and began to develop a plan for the organization to conduct fundraising events and activities. The first and very successful activity implemented was a "**50/50 Raffle.**" IWSA member families were encouraged to sell tickets, and during WAGR Weekend 2011 in Detroit a winner was drawn. The total monies raised as a result was $1750. The winning ticket holder, Steve Wright (a WAGR grandfather), wowed everyone by very generously donating his winnings back to the IWSA. This fundraising activity will be conducted again in 2012 with the winning ticket being drawn at WAGR Weekend 2012 in Maryland.

The Hefty family, IWSA Vice President Tammie and Board member Jeff, along with daughter Evie Jo, continued to creatively raise money for the IWSA. Tammie created the **Zebra Zumbathon** event, which raised more than $350. Continuing their mission from 2010, the Hefty's once again teamed up with the Milwaukee Admirals hockey team to hold the **Hockey For Hope II** fundraiser. This event combined a silent auction with the hockey game and raised more than $1200. Evie Hefty very proudly represented the IWSA and dropped the first puck to start the game!

The Dell family, IWSA Secretary Julie, husband Brian and children Madeline, Hayden, and Jaxon, continued their mission of promoting awareness of WAGR/11p Deletion Syndrome by conducting several fundraisers in 2011. Julie's creative approach to engaging family and friends through the **sale of sub sandwiches,** and their **Annual Family Donation Letter** raised more than $2000. Madeline Dell, age 11, raised $26 with a lemonade stand, and presented a check to IWSA at WAGR Weekend.

Again in 2011, IWSA Board Chair Annie Prusakiewicz presented the organization with almost $900 from the "**Casual Clothes for a Cause**" event held by the Michigan Riverview Community School District. As editor of the IWSA newsletter *WINGS*, Annie also secured a generous grant of $1200 from the March of Dimes Michigan Chapter that helped offset production and publication costs.

Board member Shari Krantz oversaw the IWSA **FLOWERPOWER** fundraiser for the second year in a row. This online sales fundraiser of flowers and bulbs netted more than $700 for the IWSA, nearly doubling the amount raised from the previous year. Many of our families across the country participated and used the opportunity to promote awareness of WAGR/11p Deletion Syndrome in their local communities.

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