International WAGR Syndrome Association

2012 Annual Report

International WAGR Syndrome Association

PO Box 392 Allen Park, Michigan 48101

WWW.WAGR.ORG

The mission of the IWSA 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 IWSA is led by a group of volunteers who strive to fulfill the mission of the IWSA. As the mission is pursued, we continue to become connected to more and more families around the world who are affected by WAGR/11p Deletion Syndrome. It is always a joy to receive feedback regarding how we have been able to help families connect, cope and care for one another. But there is always work to be done, so we continue to enhance and expand in several key areas: our knowledge, through conferences and webinars; our communications, through website updates and upgrades, and increased activity on our social networks; and our fundraising, through innovative thinking, family participation, and new partnerships with others who share the same vision.

Amy, Jenna, and Sarah get a long awaited group hug

IWSA Leadership in 2012

President
Tammie Hefty

Vice-Presidents
Jennifer Gromek and Jason Gromek

Treasurer
Nikki Hoffman, Non-Profit Consultant

Secretary
Julie Dell

Board Chairperson
Shari Krantz

Board Members
Annie Prusakiewicz, *WINGS* Newsletter Editor
Catherine Luis
Jeff Hefty
Rhonda Sena, Education Consultant
Kelly Trout, Health Consultant

Informed and Involved Leadership

The IWSA's all-volunteer leadership group participated in numerous interesting and informative meetings, workshops, conferences, and webinars in 2012. Some of the highlights include:

Future Leader, Audrey, takes an important call at WAGR Weekend 2012

Kelly Trout, IWSA Board Member and Health Consultant

• Became Alumni of the Drug Information Advocate (DIA) Fellowship Program and Co-Chaired the DIA Advocate Community 2012-2013, attended the DIA National Conference in Philadelphia, PA

• Attended the Rate Disease Symposium, NIH Webcast

• Represented the IWSA at the US Conference on Rare Diseases and Orphan Products in Washington, DC

• Represented the IWSA at the National Organization for Rare Disorders (NORD) Regional Membership Meeting in Orlando, Florida

• Attended the AHRQ Reviewers Conference in Washington, DC and served as Peer Reviewer for the publication, "Registries for Evaluating Patient Outcomes: A User's Guide, Third Edition" by the Agency for Healthcare Research and Quality, 2012

Rhonda Sena, IWSA Board Member and Education Consultant

• Attended the Wrightslaw "Special Education Law and Advocacy" by Pete Wright, Esq.

Shari Krantz, IWSA Chairperson

• Represented the IWSA (with Kelly Trout) at the Genetic Alliance "Genetic Testing and Data Management Summit: Improving Health Outcomes, Disease Management, and Accountable Care Delivery", Biotechnology Industry Organization, in Washington, DC

• Attended the Genetic Alliance Registry and Biobank Boot Camp in Washington, DC

• Attended the Steering Committee Meeting for the Coalition for Imaging & Bioengineering Research (CIBR) in Washington, DC

Noah and Emily deliver a persuasive appeal to their parents to allow them to stay longer at WAGR Weekend 2012

Tammie Hefty, IWSA President

• Graduated from "Partners in Policymaking" in the state of Wisconsin, which involved six rigorous weekend-long training sessions to learn about disability legislation and how to best impact the course of legislation regarding disability issues

• Launched the Facebook support group for the IWSA as a new means to reach out to families around the world-it has connected 58 different families from 10 different countries

• Led efforts to allow families around the world to attend WAGR Weekend virtually, via live Webcast, and through social media including Facebook and Twitter

IWSA Chosen to Participate in GRDR

In April 2012, the IWSA was one of thirty rare disease organizations chosen to participate in a pilot project by the Office of Rare Diseases Research at the National Institutes of Health. The project is called the Global Rare Diseases Patient Registry and Data Repository (GRDR).

WAGR Weekend 2012

The GRDR is designed to demonstrate how online registries of patient data can facilitate research and accelerate the development of treatment for rare disorders. The GRDR project is also intended to demonstrate how patient advocacy organizations, even those groups as small as the IWSA, can create and manage patient registries.

The IWSA Patient Registry will allow patients and/or caregivers to provide information on symptoms, diagnoses, and medical history in a secure online site. All information will be "de-identified", meaning it will be stripped of information that would allow identification of the contributor. Participants will be able to see summarized data, such as the number of other participants reporting a given diagnosis, but no specific patient identifying information.

During 2012, members of the IWSA Registry team learned about a wide variety of topics, ranging from standards for data collection and management to patient privacy and confidentiality. In October, IWSA submitted its application for Institutional Review Board (IRB) approval. IRBs evaluate research projects and insure that ethical and privacy standards are met. The IWSA received approval from Chesapeake IRB, which will also provide ongoing oversight.


Listening to presentations at WAGR Weekend 2012

Researchers wishing to study WAGR Syndrome/11p Deletion Syndrome may contact the IWSA, and if approved, Registry participants may choose whether to contribute their data but will be under no obligation to do so.

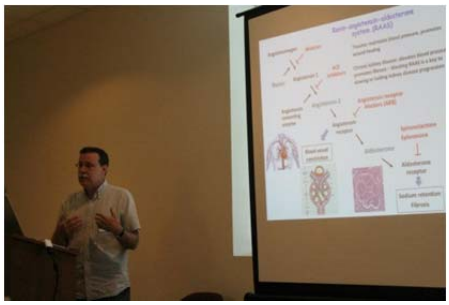
The IWSA Patient Registry Team is currently building the website that will house the Registry and tentatively plans to launch later this year.

Update on the WAGR Syndrome, 11p Deletion and Aniridia Study at the National Institutes of Health

Nicholas at the NIH with Wadih Zein, MD

We are happy to report that 2012 has brought continued progress for the genotype-phenotype study at the National Institutes of Health in Bethesda, MD. The goal of this study is to learn how people's particular genetic differences (genotype) affect their clinical symptoms (phenotype). To date, 44 children and adults with WAGR/11p deletion syndrome have come to the NIH to participate in this study.

In 2012, the WAGR study research team also presented two abstracts at scientific meetings. The first abstract, "Pineal Hypoplasia and Decreased Melatonin Secretion in Patients with *PAX6* Haploinsufficiency due to WAGR Syndrome or *PAX6* Mutations, " was presented at the 94th Annual Meeting of the Endocrine Society in Houston, TX, June 23-26, 2012. This abstract reports that people with *PAX6* deletions or mutations have underdeveloped pineal glands on brain magnetic resonance imaging, lower levels of a hormone made by this gland, melatonin (which is important for circadian rhythm and sleep), and a trend toward greater sleep disturbance. We are delighted to report that this abstract generated much interest among the scientists and physicians at the meeting and was selected as a winner in the Endocrine Society Presidential Poster Competition. The second abstract, "Measuring Autism Symptoms and Diagnosis in Select Rare Genetic Diseases," was presented at the 3rd Conference on Clinical Research for Rare Diseases in Rockville, MD, October 2, 2012. This abstract describes the potential pitfalls in using typical screening tools for autism diagnosis in patients with significant cognitive impairment, concurrent medical illness, and/or sensory deficits, such as low vision, and advocates modified approaches based on our experience working with patients with genetic disorders, including WAGR/11p deletion syndrome.



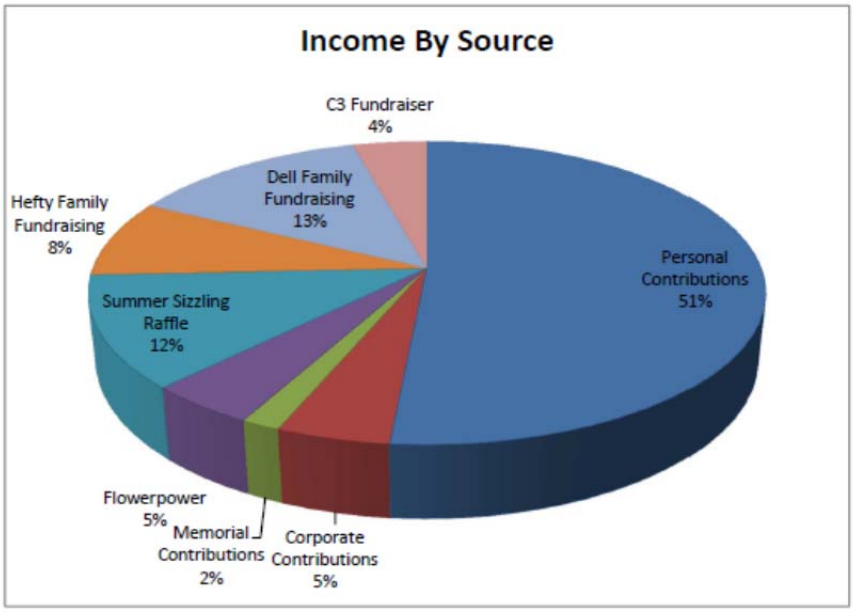
Jeffrey Kopp, MD speaking at WAGR Weekend 2012

Also in 2012, our team has been working on starting 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. We are grateful to the families who assisted us in July 2012 to pilot a chicken nuggets test which will become a very useful part of our proposed clinical trial. Progress in launching the trial has unfortunately been stalled due to the ongoing federal budget issues, but we hope to begin the study as soon as possible.

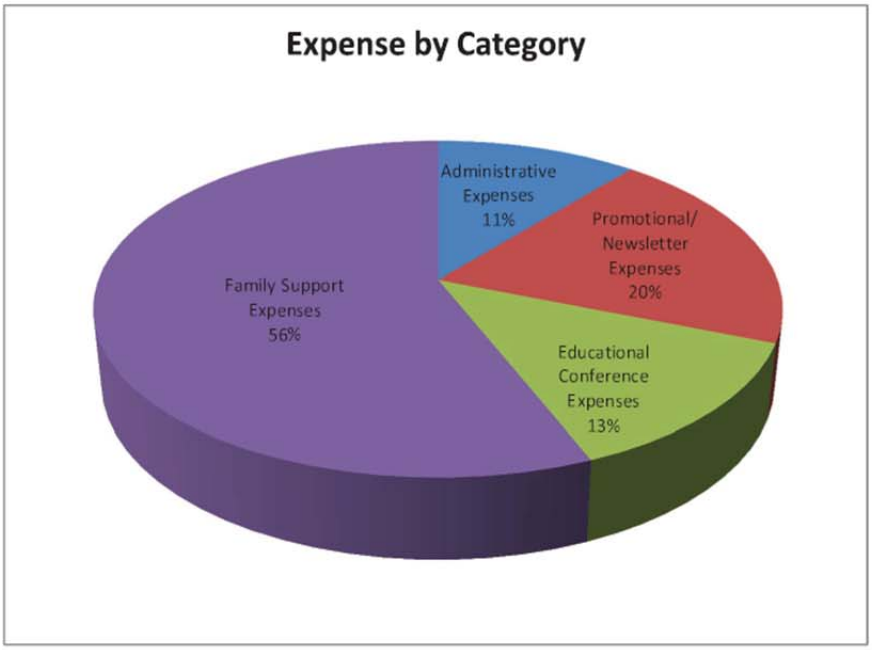
2012 Income Statement for the IWSA

International WAGR Syndrome Association
2012 Income Statement

Income		
Contributions		
Personal Contributions	$ 9,879.16	
Corporate Contributions	900.70	
Memorial Contributions	310.00	
Other - Goodsearch	7.72	
Total Contributions		$ 11,097.58
Fundraising Income		
Flowerpower	866.00	
Summer Sizzling Raffle	2,305.00	
Hefty Family Fundraising	1,576.00	
Dell Family Fundraising	2,566.00	
CS Fundraiser	790.30	
Total Fundraising Income		8,103.30
Total Income		19,200.88
Expenses		
Administrative Expenses		
Bank Charges	1.00	
Board of Directors Meetings	1,245.44	
Technology - Acct. Software	280.35	
Legal & Professional Fees	105.95	
Miscellaneous	194.52	
Office Expenses	75.00	
Organizational Fees	45.00	
Total Administrative Expenses		1,947.26
Promotional/Newsletters Expense		
Constant Contact	210.00	
Technology - website	177.49	
Mailings/Postage Business	1,110.35	
Stationery & Printing	572.00	
Newsletter	1,410.00	
Total Promotional/Newsletters Expense		3,479.84
Educational Conference Expenses		
Conference Fees	250.00	
Travel - Conference	2,014.12	
Total Educational Conference Expense		2,264.12
Family Support Expense		
Travel Costs	241.59	
Gifts	226.71	
WAGR Weekend		
Miscellaneous	3,400.00	
Family Assistance Scholarship	2,658.75	
Meals	3,326.87	
Total WAGR Weekend	9,385.62	
Total Family Support Expense		9,853.92
Total Expenses		17,545.14
Net Income		$ 1,655.74



Sources of Income and Expenses by Category



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