

Listening To You

The Newsletter for AGRE Families

Autism Genetic Resource Exchange

AGRE

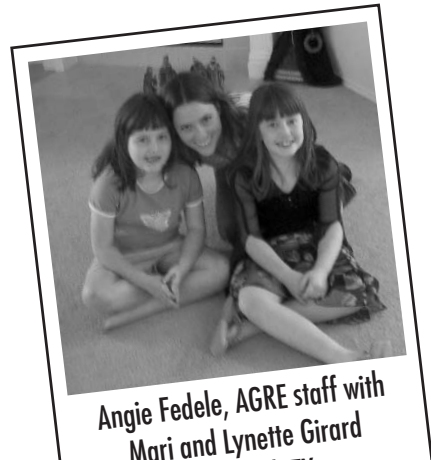
info@familyagre.org
www.familyagre.org
866-612-2473

CAN

Cure Autism Now
info@cureautismnow.org
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323-549-0500



Tyler Toth, Brienne Cohen,
and Zachery Toth
Columbus, OH



Angie Fedele, AGRE staff with
Mari and Lynette Girard
Mansfield, TX



Jacob Toth, Columbus, OH
with Brianne Cohen, AGRE staff



Sunshine, Mickey, and Eric Taylor
Buffalo Gap, TX



Hunter Toedman
Aptos, CA



Special Interest Articles:

2004 Summary Report

2005: Looking Ahead

Family Matters

Science News

Cure Autism Now

Individual Highlights:

Best 2004 Staff Memories

From the Director

New AGRE Staff

A Successful Year for AGRE

AGRE has expanded the collection of clinical data and genetic material to include over 600 families. As a result, AGRE is now the world's largest autism gene bank that is made available to researchers

throughout the world. Ten years ago, a family collection for the genetic study of autism was little more than a dream of the family members and scientists who started Cure Autism Now. Their goal has always been to

help scientists find the genes that would lead to new therapies, and perhaps even a cure, for individuals diagnosed with autism spectrum disorders.

Continued on page 3



Martine Ferguson, Clara Lajonchere, Catherine Stranieri
Paris, France

2004 Highlight:

In November, Clara Lajonchere, Ph.D., AGRE Program Director, was invited to attend a conference on autism genetics by *Fondation Autisme*, a parent advocacy group in Paris, France. The conference was sponsored by *IntegraGen*, a French biotech firm that has purchased AGRE samples in the past. CAN and AGRE are excited about potential partnerships with organizations like *Fondation Autisme* in Europe and abroad. ■

2005: Looking Ahead

Over the last eight years AGRE's primary mission has been to support autism researchers in their investigations. As a result, researchers have made remarkable

progress in identifying several chromosomal regions associated with the disorder. Researchers now know that autism is not caused by one gene, but believe it is the result of the interaction

of many genes. Because of the complexity of the disorder and differences in individual symptom profiles, AGRE will continue to spearhead autism research.

AGRE's Key Objectives for 2005:

- Continue commitment to recruit families with two or more children diagnosed with autism, PDD, or Asperger's Disorder. Recruiting additional families will build the resource to its largest capacity, thereby increasing adequate statistical power for genetic analysis.
- Increase public and private partnerships. These collaborations will position AGRE to further support new investigators, aid in rapid replication of new findings, and facilitate the immediate application of the latest tools and technologies.
- Expand data collection to broaden the data available to researchers. AGRE hopes to include data on social responsiveness, speech and language development, and behavior patterns.
- Create a "Library of Genetic Findings" which will serve as a portal for autism researchers worldwide. Creating a centralized repository of the latest findings in genetics, will aid researchers interested in the field to ask the right questions and avoid duplication of efforts. ■



Tom Ferguson
Paris, France

AGRE Autism Genetic Resource Exchange
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Clara Lajonchere, Ph.D.
AGRE Program Director

member, I can only respond "Why not?" First of all, I love children and want their lives to be as full as possible. As a scientist, I am fascinated with how the brain works, and as a researcher, I want to understand how an illness like autism can happen. As an optimist, I know that we can find the cure. "Impossible" is a word found in the dictionary of fools. Therefore, I am certain that solving this mystery is not impossible.

AGRE's contribution to the research conducted by the scientific community continues to be remarkable. While we are grateful to our researchers for helping accelerate the pace of science, we must recognize that our families are the real heroes. You remain our partners and our greatest stakeholders, the heart and soul of our program. Without you, we could not have such an impact.

When asked why I work tirelessly to find a cure for autism given that I don't have an affected child or family

I have seen and felt the passion of our families, our board members and our staff, and am convinced this will continue to drive our organization further into its mission to "Cure Autism Now". To be a part of this has been an honor for me, and I remain dedicated to our children, to autism, and to the advancement of science. At the end each day, I know we will do our part to what will someday soon be a breakthrough in our understanding of and treatment for autism. It is as simple as that! ■

AGRE Steering Committee:

Chairperson: Maja Bucan, Ph.D.

**Chief Scientific Advisor:
Dan Geschwind, M.D., Ph.D.**

W. Ted Brown, M.D., Ph.D., F.A.C.M.G.

Joseph Buxbaum, Ph.D.

Rita Cantor, Ph.D.

John Constantino, M.D.

T. Conrad Gilliam, Ph.D.

Martha Herbert, M.D., Ph.D.

David Ledbetter, Ph.D.

Stanley F. Nelson, M.D.

Gerard D. Schellenberg, Ph.D.

Carol Samango-Sprouse, Ed.D.

Matt State, M.D., Ph.D.

Rudolph E. Tanzi, Ph.D.

Family Recruitment

If you know of an autism support group, family conference or mailing list that would support AGRE's recruitment in your area, contact Marianne Toedtman at 888-288-4762 or mt@agre.org

Stay in Touch!

Update Your Family's Contact Information.
Let us know if you've changed your address, added a new email, or a cell phone number!
Use our online form at www.familyagre.org or call 888-288-4762.

AGRE was created by CURE AUTISM NOW to advance genetic research in autism spectrum disorders. DNA samples and clinical data are obtained from families that have more than one member diagnosed with autism, Pervasive Developmental Disorder (PDD) or Asperger's Syndrome. Data is immediately made available to qualified researchers.



Coming to Terms with Autism

by Dave Piszcz

Parenting is a test of one's life skills at all times, but the standards for passing those tests become higher if a disabled child is involved.

In Ken Joondeph's case, both of his sons - Joseph, 12, and Tyler, 10 - have been diagnosed with autism. Daily life is an intense, sometimes frustrating, yet often rewarding experience, he said.

Tyler, whose autism is much more profound than that of his older brother, has made significant progress, given that he inhabits a much more closed internal world. Tyler remains essentially nonverbal, but can communicate through the use of pictures and American sign language, although his spoken vocabulary is on the rise.

Joseph, a major movie fan who often communicated by bits of dialogue from his favorite films, is speaking in a more straight-ahead

fashion. "His communication is less 'scripting' now. He's sometimes in his own alternative reality and he needs to be brought back," Joondeph said. "That's becoming less and less. Joseph is able to ask things like 'How was your day?' or 'What can we have for dinner? Can I help?' He's able to reflect on what he did during his school day."

A typical day at the Joondeph household begins at about 5:30 a.m. when Joondeph wakes up, has breakfast and then rousts Joseph and Tyler out for showers and food. Home support workers come by at 6:30 a.m., which is Joondeph's usual leaving time when he's working in the classrooms.

He described the atmosphere of the household as similar to that of a life-skills classroom.

"The baby steps are the best," Joondeph said. "When you see a

child able to overcome one step and move on to the next, with increasing speed, they're getting it. That's one of the pleasures of being a special ed teacher, because you really see the results. Teachers do learn from their students, too. I really feel that being a special ed teacher with two special needs kids, I'm in a very unique position to constantly learn, to constantly re-tune and refine my skills."

"I will always be involved with my children's life," he said. "They will always need staff, maybe not 24 hours a day, but I would always want to have some kind of input in their lives. As I get older and older, I'll have some kind of contract with Group Home Foundation or another service provider to staff the house and run it."

Dealing with two autistic children is a full-time job and then some, Joondeph admitted.

This article was edited for length. Find the complete article online at: www.courierpub.com/couriergazette ■



Joondeph Family
Belfast, ME

New AGRE Staff



Debra Busdiecker, AGRE Research Associate, graduated from Texas Christian University (TCU) in Fort Worth, TX. As a student she worked for the Developmental Research Lab at TCU in the Hope Connection, a camp for special needs adopted children. In 2004 she moved to Los Angeles, and joined AGRE in September, as a Research Associate. Her career plans include going to graduate school to study developmental psychology and continuing her work with children with special needs.



Linda Clark, Phlebotomist, completed Phlebotomy and EKG training at Illinois Medical Training Center, Chicago, IL in 1982 and became a California Certified Phlebotomist in May, 2002. Linda joined AGRE in March, 2004.



Cindy Huang, Diagnostic Clinician, graduated from University of California, Los Angeles (UCLA) with a Bachelor's degree in Psychology. She worked as a research assistant at the UCLA Autism Clinic from 2000-2003. She joined AGRE in August 2003. Her duties include administering the Autism Diagnostic Interview - Revised (ADI-R) to parents, and conducting the Autism Diagnostic Observation Schedule (ADOS) with children in their homes.



Cure Autism Now Announces New Science Director



Sophia A. Colamarino, Ph.D.
Science Director

Dr. Colamarino comes to Cure Autism Now with exceptional research credentials and an intense interest in both biology and psychology. She will be responsible for guiding Cure Autism Now's science program in association with the Scientific Advisory Board and Scientific Review Council as well as liaising with the AGRE Steering Committee.

Dr. Colamarino's background in developmental biology, particularly extensive studies in brain development and regeneration, make her a welcome addition to the autism field. Her vision for Cure Autism Now is to see it take a leadership role in advancing what is known about the biology of autism and ultimately defining its cause on a cellular and molecular level. ■

WEB RESOURCES



Cure Autism Now - Are you CONNECTED? Be sure to sign up for CAN's eNewsletter, *Connections*. Click on "get eNewsletter" to receive news and updates in one convenient biweekly email.

www.cureautismnow.org

Autism Tissue Program - provides scientists interested in studying autism access to human brain tissue. A national outreach program for families makes them aware of the need for this gift of hope.

www.memoriesofhope.org

First Signs - educates parents and physicians about the early warning signs of autism and other developmental disorders in early childhood.

www.firstsigns.org

National Institute of Mental Health (NIMH) - gives the latest government updates on research, health, and funding information on autism.

www.nimh.nih.gov/nimhhome

Future Horizons - autism and Asperger's Syndrome book publishing. The site also lists autism and Asperger's Syndrome national conferences.

www.futurehorizons-autism.com

Cure Autism Now Chapters

Chapters play a crucial role in helping Cure Autism Now to raise funds for autism research and to increase awareness of autism and related disorders in local communities.

You can join one of the thirteen chapters around the country:

- Atlanta Chapter, Georgia
- Chicago Chapter, Illinois
- Hawaii Chapter
- Houston Chapter, Texas
- Greater Los Angeles Chapter, California
- New York City Chapter, New York
- Orange County Chapter, California
- Orlando Chapter, Florida
- Pacific Northwest Chapter, Washington
- Philadelphia, Pennsylvania & Southern New Jersey Chapter
- Mid-Atlantic Chapter, Maryland
- San Francisco Bay Area Chapter, California
- Tri-State Chapter, New Jersey

WALK NOW Goes Coast to Coast in 2005

WALK NOW is Cure Autism Now's grassroots fundraising and awareness initiative uniting thousands of parents, children and families in a fun, friendly, empowering environment. The Walk is a 5K (just over 3 miles) with lots of water, food and fun along the route. And each WALK NOW event includes a Resource Fair. Parents can meet a variety of autism service providers while kids enjoy arts & crafts, moon bounces and other fun activities.

WALK NOW events will be held in thirteen cities this year. Join us in walking toward a common goal—to find the causes, effective treatments and a cure for autism.

- Atlanta, April 2005
- Baltimore/Washington, September 2005
- Boston, June 2005
- Chicago, May 22, 2005
- Honolulu, March 12, 2005
- Houston, November 2005
- Los Angeles, April 2005
- New York City, September 2005
- Orlando, June 2005
- Philadelphia, October 2005
- San Diego, November 2005
- San Francisco, October 2005
- Seattle, September 2005



For more information or to register, visit www.walknow.org. Questions? Call us at (888) 8-AUTISM or email us at: walknow@cureautismnow.org.

Family Recruitment

In 2004 an increasing number of families registered to participate in AGRE, with a majority of new families registering through the website. Staff attended many family autism conferences to increase awareness and participation in AGRE. Our goal is to recruit 300 new families in 2005.

Family Satisfaction

Families who participated in AGRE are asked to complete a satisfaction survey designed to improve communication and better understand families' needs. In 2004 responses from 140 families highlight the overall positive view families have of AGRE, while pointing out that feedback is a priority for families, who want to know research updates and important scientific findings.

Collaborations

This has been the area of greatest growth for AGRE. The best way for AGRE to stand out as the world's largest gene bank for autism research is to maximize resources through collaborations. In this way, we are able to increase both the depth and breadth of the collection.

- Washington University in St. Louis—Social Responsiveness Scale -completed
- Lipomics Technologies, Inc.—32 families completed, 8 remaining
- UCLA - Brain Imaging Study—in progress
- Stanford University—Twin Study—funded
- Center for Inherited Disease Research (CIDR)—Phase 1 completed
- Perlegen Sciences—in progress
- Autism Genome Project (AGP)—in progress
- Southwest Autism Research and Resource Center (SARRC)—co-recruitment

Conferences

AGRE was an exhibitor at 7 autism conferences. Members of the AGRE team were featured speakers and/or presented posters at state and local conferences including:

- IMFAR, May, 2004-C. Lajonchere
- Texas State Autism Conference, Sept. 2004—C. Lajonchere/S.J. Spence

- 35th National ASA Conference in July 2004—C. Lajonchere/S.J. Spence
- Siever Conference in New York City, November 2004—M. Lutz

Media Publications

Marianne Toedtman, Outreach and Resource Manager, submitted stories for AGRE that were featured in the Autism Digest Magazine, May-June 2004, and Exceptional Parent Magazine, November 2004. AGRE will also be featured in Spectrum Magazine early 2005. Increased visibility in print materials has increased the number of families interested in the AGRE collection.

AUTISM Researchers and Families Combine Efforts to Understand Autism

By Marissa Taczinski RN, MS

Families make the critical difference
The Autism Research Center (ARC) at the University of Washington is a leading center for autism research. The center's research focuses on understanding the genetic and environmental factors that lead to autism. The center's research is focused on understanding the genetic and environmental factors that lead to autism. The center's research is focused on understanding the genetic and environmental factors that lead to autism.



Exceptional Parent Magazine 10/04
To read the article visit:
www.eparent.com

Serving the Research Community

AGRE's 2004 research activities include:

- Researchers can access information from over 600 families for use in their investigations.
- There are 135 AGRE-approved researchers who have access to study families in the AGRE collection.
- AGRE samples were cited in more than 17 articles in major medical and science journals, in 2004 alone, and in 37 since the first publication in 2001.

AGRE Funding

Support from Cure Autism Now as well as additional funding sources serve to further enhanced the AGRE resource through the following partnerships:

- NIMH grant funding through collaborations at UCLA
- Dansko Foundation
- Quest Diagnostics

AGRE's Institutional Review Board (IRB) Oversight

AGRE is committed to ensuring the protection and privacy of all our research participants. Any research study involving human subjects must be overseen by an Institutional Review Board (IRB), an outside agency that reviews and monitors biomedical research with human subjects. The staff at AGRE takes every measure to remain in compliance with state and federal regulations as they pertain to our families. This means that all consent forms, assessment instruments, and recruitment documents must receive the approval of the IRB before we are able to use them with our families.

AGRE is primarily overseen by the Western Institutional Review Board (WIRB), one of the oldest and most experienced independent IRBs in the United States. WIRB has provided review services for the research of more than 10,000 investigators in over 30 countries, and in all 50 United States. In addition, AGRE must obtain approval from the IRB at the University of California, Los Angeles (UCLA), because we are receiving NIMH funding through our collaborators at UCLA, Dr. Daniel Geschwind and colleagues.

In order for us to continue collecting data from families, AGRE must obtain updated approval from WIRB and UCLA each year. Without approval from these independent agencies, AGRE's recruitment and data collection activities would come to an end.

The AGRE families are both our partners and our greatest stakeholders, and we are committed to the protection of their privacy rights. Our goal is to ensure that the risks of scientific advancement never outweigh the value and well-being of our families. ■

Does your family or a family you know have two or more children diagnosed with autism, PDD or Asperger's Disorder? This is an opportunity for families to contribute to scientific research. Contact AGRE at 888-AUTISM-2 (288-4762) or email: mt@agre.org

"Having the blood draw done in the comfort of my own home was so much easier on my children, who hate needles of any sort. That the procedure went so smoothly is testimony to Linda's (phlebotomist) great people skills and clinical expertise when it comes to her job".

AGRE Family
Arden Hills, MN

Borgnesser Family

Leonardo, NJ

From the moment our son Kevin was born, I suspected there was a problem. My best friend, who has experience with autism in her own family, confirmed my concerns by pointing out her own observations of Kevin. Several doctors assured me that Kevin would catch up. But after what seemed like an eternity, Kevin was diagnosed with autism at the age of two.

My husband and I immediately began early intervention in our home and quickly saw signs of progress. We also attended autism workshops and learned strategies to educate our son.

On Kevin's third birthday he started going to school. We were very fortunate to have Kevin attend a school for autism that shared our goals and dreams for his future. His progress continued to amaze us.

During this time, our middle son, Paul, was diagnosed with Asperger's Syndrome.



Paul Jr, Kevin and Samantha Borgnessar

Our experience has been very positive. The professionals that have come into our home have been knowledgeable, compassionate, patient, and kind. My children view the experience as a game and have enjoyed themselves. Our family will continue to be part of AGRE as long as we can provide useful information for the study.

As Samantha, our 12 year old daughter, pursues her dream of finding a cure for autism, our family will continue to encourage our children's hopes for the future. Paul's dream is to join the NFL and Kevin aspires to become an art teacher – and we believe there are no limits to what they can do!

Thank you so much for working to build better tomorrows for families like ours, and the ever-growing autistic population. ■



Stapleton Family
San Juan Capistrano, CA

Toth Family

Columbus, OH

They say that "God only gives special children to very special people," which makes me laugh and think that my husband and I are certainly blessed.

We have four children: Erin is 21, Tyler 9, Zachery 7, and Jacob 5. In 2001, Zachery and Jacob were both diagnosed with Autism Spectrum Disorder within six months of each other. Life as I knew it was over, but a new life emerged when I found Dr. Rebecca Morrison and The Children's Center for Developmental Enrichment (CCSE) in Columbus, Ohio. The support from families and friends I have met there has meant so much to me.

The CCDE's mission is to provide services for families and their children with autism-related disorders. They believe all children should be viewed in terms of their strength and capacity for learning, as well as their ability to participate in the community.

All children benefit from belonging to a group of their peers. It is in childhood that we learn to compromise and care for others. Tolerance for individual differences is developed and celebrated, while diverse learners are encouraged and supported to reach their potential.

CCDE provides affordable and accessible educational services for children with ASD in fully inclusive classrooms with one-on-one assistance. The school supports families of children with ASD by providing a range of resources and mental health services. CCDE promotes community awareness and collaboration in the fight to defeat the impact of ASD on the individual's successful assimilation in the community.

This school is on the cutting edge and it has been wonderful, not only for Jacob and Zachery, but also for my son Tyler who loves being a leader and a helper for his classmates. He has learned incredible compassion and

goodness, and each day proves his love for his classmates and his younger brothers at home.

I thought how wonderful it would be if my family could contribute something to uncover the causes of autism and, hopefully, find a cure. My husband asked me why we would want to participate in this study. I told him that if we could save one family the heartache of autism, it would be worth it.

We are so proud that we could participate in the multiplex family study conducted by the Autism Genetic Research Exchange. I hope our participation helps. ■



Zachery Toth



Jacob Toth

"I thought how wonderful it would be if my family could contribute something to uncover the causes of autism and, hopefully, find a cure. My husband asked me why we would want to participate in this study. I told him that if we could save one family the heartache of autism, it would be worth it."

Cynthia Toth

Proteomic Studies Identified a Single Nucleotide Polymorphism in Glyoxalase I as Autism Susceptibility Factor

Mohammed A. Junaid, Dagmar Kowal, Madhabi Barua, Premila S. Pullarkat, Susan Sklower Brooks, and Raju K. Pullarkat

NEW RESEARCH IDENTIFIES PROTEIN LINK TO AUTISM

A report published in the November 2004 issue of the *American Journal of Medical Genetics* highlights the utility of collections like AGRE (www.agre.org), which offer scientists an unprecedented resource for the study of autism genetics.

There are many large programs underway currently focusing on the genetics of autism. While these studies are providing scientists with a genetic roadmap, none has yet identified the specific genes involved in autism. This lack of specificity led Dr. Mohammed A. Junaid and colleagues to use a different approach to examine the biology of autism. These scientists reasoned that given the differences in the brains of individuals with autism compared with controls, examining the proteins from the brain tissue of autistic individuals may lead to promising findings.

Proteins are the molecular machines that carry out the majority of work in the cells and the body. From transmitting signals in brain cells to eliminating the byproducts of biochemical reactions in the liver, proteins are the workhorses of the body. DNA is nothing more than the template for building these proteins.

Dr. Junaid and his colleagues acquired samples of donated autopsied brains from autistic (www.brainbank.org) and non-autistic individuals and extracted all the proteins from these tissue samples. This extracted set of proteins was then separated on a gel. Proteins with similar charge and size will accumulate as a spot on the gel. Interestingly, what Junaid and colleagues found was that one particular

spot, found in four of the eight autistic samples tested, was only identified in two of the nine non-autistic samples. The researchers were able to extract these proteins from the gel to uncover the protein sequence. From the protein sequence, a DNA sequence of the gene that serves as the template can be found. With a little bit of DNA sequence in hand, these scientists looked for DNA changes in these samples and identified a variation in the sequence of the gene Glyoxylase I.

Glyoxylase I is an enzyme protein responsible for scavenging and removing toxic byproducts, which cells create during normal cell processes. A dysfunction of this enzyme will result in the accumulation of toxic byproducts.

In order to determine whether this gene is involved in autism, Dr. Junaid and his colleagues examined whether the mutation causing the change in protein sequence was associated with autism in the AGRE sample compared with a set of control families. While the identified change in sequence was found in both control families and in families with autism, the proportions were very different, indicating a relationship between the changes in DNA sequence in the autism group.

Finally, the group tested how well the autism-associated protein form functioned in the human cell in comparison to the

control form. They found that there was a near 40% reduction in the efficiency in the byproduct clearance as well as the accumulation of these byproducts in the brains of autistic individuals.

While this study's results are very interesting, they warrant further investigation in a larger number of families. The identification of this gene variant and its clear effect on the removal of toxins from the body is exciting for the field and may lead researchers to a better understanding of the cellular mechanisms that are disrupted in individuals with autism. ■



*AGRE families provided samples for this scientific publication.

Science News articles contributed by Vlad Kustanovich, Ph.D., Clara Lajonchere, Ph.D., Marianne Toedtman, RN

Evidence for Sex-Specific Risk Alleles in Autism Spectrum Disorder

Jennifer L. Stone, Barry Merriman, Rita M. Cantor, Amanda L. Yonan, T. Conrad Gillman, Daniel H. Geschwind, and Stanley F. Nelson

GENDER IS A FACTOR

Autism is a disorder that affects every race, nation and creed. However, there is a clear male bias in susceptibility to autism and Pervasive Developmental Disorders. Recent reviews have indicated that males are four to ten times more likely to be affected with autism than females. Interestingly, other neurodevelopment disorders such as attention deficit disorder and dyslexia reflect a similar male: female bias.

As a result, many researchers expected to identify susceptibility genes on the sex chromosomes. The first genome scans left researchers baffled because these signals were not evident. Researchers found it difficult to reconcile why there would be a bias in affected males that did not involve the sex chromosomes.

Scientists reasoned that one possible explanation for this phenomenon was that genes contributing to the susceptibility of

autism are modified by sex hormones. In a report published in the September 2004 issue of the American Journal of Human Genetics, a group of scientists at UCLA and Columbia University identified a clever strategy to discover genes modified by sex-specific factors.

Jennifer Stone and colleagues used the existing genome scan data performed on AGRE families in 2003 (Yonan et al., 2003). They divided families into two groups: those containing only males affected with autism, and those containing both affected males and females. The rationale for this division was based on the idea that some autism susceptibility genes will be influenced by sex-specific risk factors. One can assume that the genes of male-only families are enriched for having male risk factors, while those of families containing both males and females have risk factors that affect both sexes.

ABOUT THE GENOME SCAN

A genome scan is carried out by performing many genetic tests on every available family member in a large collection of families (e.g., AGRE). The genetic tests determine which form or variant of a genetic marker (a piece of DNA that is used to tag specific areas of the genome) is passed from parents to children. When the forms of all the genetic markers are tabulated, statistical analyses can be made of this information to determine if certain areas in the DNA are shared among individuals with autism. This sharing is the foundation of how human geneticists identify regions in the DNA that play a role in diseases.

CHROMOSOME 17

Amazingly, a genetic region on chromosome 17 identified in the original genome scan was seen as highly shared among affected individuals in the male-only families. More importantly, this trend toward higher sharing was much stronger than in the original genome scan despite the reduction in the number of families

examined, because the families were divided into two nearly equal groups (58% male only families; 42% female containing families). Interestingly, when the genome scan was performed on female containing families, the chromosome 17 region did not indicate any sharing, suggesting that all the statistical signals seen in the original scan were generated almost exclusively from the male-only families.

BEYOND CHROMOSOME 17

Aside from the chromosome 17 region, 24 other genetic regions were found to be different between this genome scan and the original 2003 scan, but none reached the significance of the region found on chromosome 17. Some scientists may question whether the division of families into male-only and female-containing families was arbitrary and therefore question the meaning of the findings. To avoid this type of criticism, the scientists developed a method whereby all families in the initial analyses were randomly assigned to two groups, regardless of whether they were an affected male-only family or a mixed male-female family. This process was repeated 1000 times to try to replicate the amount of DNA sharing at particular genetic regions without the sex-specific bias. This procedure also served to create a baseline of what could occur by chance. Interestingly, the replicates did not produce any signals close to that identified on the chromosome 17 region in the sex-specific samples.

CONCLUSION

Because of its identification as a region likely to harbor susceptibility genes in the original genome scan, the region on chromosome 17 is actively being examined as a potential area of future research. Several groups have already examined the serotonin transporter gene also found in the chromosome 17 region. However, the evidence is not entirely conclusive, and a more thorough examination of this candidate genes as well as the surrounding region is being carried out. ■

Montoya Family

Oaklawn, IL

My husband, John, and I have three children: Jay, Matt and Ryan. Matt and Ryan are both autistic. Matt, who is now 10, was diagnosed just before his fifth birthday.

I sensed there was something wrong with Matt. He repeated everything, made little eye contact, and obsessively lined things up. By the time Matt started school, he was nonverbal. It was the school district professionals that suggested to us that our son had autism.

Matt is now considered high functioning, but not Asperger by any means. He has made tremendous improvement in school, at home, and in the community. Matt likes to swim, ride his bike, go to the zoo, watch videos, and help me bake.

Just as I thought I understood autism, Ryan came along. Ryan met all his first-year milestones, but still we watched for the signs. Unfortunately, we saw them. The same professional group that diagnosed Matt evaluated Ryan.

As we were gluing the shattered pieces of our lives back together, our hearts were broken again. This time, I wasn't sure I was strong enough to pick up the pieces. But I at least knew where to start. Our school district helped immensely. Ryan was placed in an early intervention program for one year and now is in a program for autistic children through our school district. Ryan enjoys videos, singing, dancing, swinging at the park, and playing on the computer. He is doing well in school.

Even though both boys are autistic, they couldn't be more different. Matt is verbal, Ryan is not. Matt has many behavior issues and Ryan is very mellow and quiet. Matt is skinny as a rail, and Ryan is a "little brick house", as my husband puts it.

When I compare my sons to normal children, I see two brothers who love to laugh, be tickled, and play with each other. Their joy is contagious and it comforts me to see that, in so many ways, we are normal. Sometimes I see my sons show a range of emotions - from scared to affectionate, to fierce and determined. I see them discovering the world and finding their place in it.

What I have learned is to see children first before I see children with autism. I realize I am exactly where I'm supposed to be, and with the children I was destined to have in my life.

I became involved with AGRE in the hope that working together, we will find answers and cures for autism that will help children lead the full lives their parent's dream of. ■



Ryan and Matthew Montoya

Robb Family

Baskingridge, NJ

My husband and I have been blessed with 5 beautiful children. Taylor 9, Dougie 7, Jaxon 5, Peyton 4, and Mia Grace 2.

Both of our boys have been diagnosed with ASD: Dougie with PDD/NOS/ADHD and Jaxon with autism.

Dougie was diagnosed early, when he was about 18-months-old. Jaxon was diagnosed when he was just over two-years-old. But it wasn't until Jaxon was three that he was diagnosed with autism. As Jaxon gets older, the stereotypical behaviors associated with autism become more pronounced.

Recently, we moved from our home of four years. We are now in a new district that offers an incredible autistic program for Jaxon. Dougie also is in a great program and has been mainstreamed into first grade with an aide. The district also offers a terrific education for our girls.

As parents, our challenges are difficult. We are raising two children with special needs, as well as three other children who have their own individual needs. Yet, every day we are strengthened by our faith, which allows us to see beyond the disabilities and the troublesome behavior.

We have learned that we cannot take anything for granted. Everything is a gift from above and our boys are

truly blessings - not only to us, but also to everyone who knows them. Dougie and Jaxon are precious gifts with beautiful spirits and awesome talents.

We decided to join AGRE after attending the "Walk Now Philadelphia 2004." It inspired us to do our part to help find a cure. After all, our boys are included in the 1 in 166 statistic. We couldn't sit by without taking action any longer.

Like any family dealing with autism, we want to find the cause and the cure during our children's lifetime - the sooner the better! That is why we want to help in the effort to "hurry science".

We are grateful for the support, understanding and efforts of AGRE and Cure Autism Now, and look forward to working with both organizations as long as it takes to find a cure. ■



Dougie, Mia, Peyton, Jaxon, and Taylor Robb

O'Donnell Family

Austin, TX

Earlier this year, 10-year-old Deirdre O'Donnell penned a poem in honor of her sister with autism, Caitlin.



O'Donnell Family

'My Autistic Sister'

Autism silences your beautiful voice
From telling me what you are feeling
What causes you to laugh
When I don't get the joke
To the tears you shed
When I don't know your pain

Or your favorite song and
When you hug me when you

Fear the thunderstorms
Not knowing what torments you so often
Keeps me from comforting you.

Although autism silences your voice
I see love in your gleaming smile.
Even though I don't get your joke

I find myself laughing myself to tears
Just to see joy emblazoned on your face.
You are my sister

And that is what defines you
Not AUTISM.



*AGRE families provided samples for this scientific publication.



"In May of this year, we made a big push to change the Institutional Review Board (IRB) that protects the rights of our families. One of my best moments was finally getting approval from our new IRB, after many months of work (and lots of coffee)!"
Janet B. Miller,
Clinical Director



"When I visited a family's home, the child thought I was Pocahontas. What a wonderful compliment!"
Brianne Cohen,
Clinical Research Associate



"It was the day a researcher told me his research simply would not have been possible if it wasn't for the AGRE resource".
Vlad Kustanovich,
Researcher Liaison



"Being promoted to data analyst. By continually upgrading our database and auditing our data, I know that our researchers have access to the best data available".
Jocelyn Furr,
Data Analyst



"The day I got my job. I was so excited to join the team".
Debra Busdiecker,
Research Associate



"It was during an autism interview with a non-verbal child. During the assessment, the boy said to his family, 'Happy Birthday.' It was quite an emotional experience for all of us. And I am deeply touched to have been a part of that profound moment in their lives".
Angelina Fedele,
Senior Clinical Research Associate



"My mother sent me a newspaper article from Austin, Texas about a family who had a wonderful experience when AGRE staff visited their home".
Nancy Hart,
Data Projects Manager

**Wishing you
 & your family
 a new year
 filled with
 wonder, joy,
 & hope.**

From The AGRE Staff



"When I heard that AGRE had been cited in seventeen scientific publications".
Marianne Toedtman, Outreach & Resource Manager



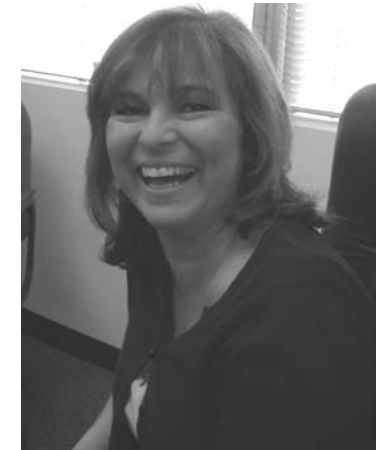
"I saw a parent in my office at UCLA. While taking the family history, I realized they had two children with autism. I was about to discuss the AGRE research project with them. But before I could utter a single word, the parents asked me if I knew about a wonderful research program for families with autism they were involved with - 'It's called AGRE!'"
Sarah J. Spence,
Medical Director



"Seeing tears of hope in the eyes of parents after I've given a talk on the AGRE resource".
Clara M. Lajonchere,
Program Director



"I was staffing the AGRE booth during a family conference and a parent asked me: Has anyone told you thank you today? I want to personally thank you for being here for us. It really means a lot".
Tiffany Torigoe,
Family Recruitment Specialist



"It was the day we asked a family how AGRE could improve my visit to their home. They said, 'How can you improve a vampire's visit?'" (Smile)
Dani Ocampo,
Phlebotomist



"The AGRE families' enthusiasm to participate in the Lipomics collaboration, especially since it involved an additional blood draw for selected family members".
Marin Lutz,
Collaborations Manager