



AGRE FAMILY NEWSLETTER

Autism Genetic Resource Exchange

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Autism: Moving from DNA to RNA

By: Vlad Kustanovich, Ph.D., Researcher Liaison and ISAAC Project Manager

Three new publications herald a new frontier in autism research which is both timely and welcome. While there is definitely a need for studies examining the association of autism with DNA variants and genetically linked regions, DNA chip studies offer a new and powerful experimental strategy to understand the complexity of autism, and uncover the underlying causes and subtle biology that result in autism spectrum disorders.



"The AGRE repository is an invaluable resource for my research, not only for access to biological samples, but also for its well-documented collection of phenotypic data, which is crucial for interpretation of biological findings."

Valerie W. Hu, Ph.D.
Associate Professor of Biochemistry and
Molecular Biology
The George Washington University Medical Center

Three recent papers this year have used samples from the **Autism Genetic Resource Exchange (AGRE)** repository to examine changes in gene expression and RNA levels in the cells of autistic individuals. These cells are derived from the blood samples collected during the AGRE phlebotomy visit. While DNA serves as the blueprint of life, being passed down from generation to generation, RNA is the chemical messenger in the cell which is coded by the DNA template and determines which gene products are active in a particular cell or individual. The levels of a particular RNA present in a given cell determine the abundance of protein in that cell. For example, while cells in the nervous system and fat tissue may share some RNA's, the presence or abundance of specific RNA's between the two cell types will differ substantially and determine the overall focus of the cells' activities. Studying the RNA's found in cells from autistic individuals may help in identifying biomarkers for autism.

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Connections in the Community Make a Difference in Children's Future

Knoth Family, Milford, CT

My husband George and I have four amazing sons; George, Jr. (10), Justin (8), Samuel (6), and Benaiah (4). Our middle sons, Justin and Samuel, have diagnoses of autism. Introduction to the world of autism came upon us subtly. Initially we all characterized Justin as shy, temperamental and disobedient. I think in retrospect you could have called us "parents in denial". At that time, denial was easy in comparison to the thought of accepting the world of autism. We were not afforded the luxury of



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Knoth Family, Milford, CT



Clara Lajonchere, Ph.D., Program Director, attended Tufts University in Boston, MA where she received a B.S. in Psychology with a minor in the History of Medicine. She received her Ph.D. in Experimental Psychology from Washington University in St. Louis with concentrations in neuroscience and psychopathology. Clara was introduced to genetic research in 1991 when she worked on the Biological Program Project for Schizophrenia at McLean Hospital/Harvard University. Clara joined AGRE in 2003 to promote autism research and contribute to an organization that provides a collaborative approach to science. Email her at clara@agre.org

Dear Families:

Starting my fourth year as AGRE's Program Director, I continue to take great pride in the momentum we have created and the extent of our contributions to autism research.

Later this month, the release of the AGRE pedigree catalog will make DNA samples from over 830 families, the majority with two or more affected family members, available to the scientific community. This huge AGRE milestone will have tremendous impact on autism research. In August of this year, the Autism Consortium, a scientific and clinical collaboration that includes eleven Boston area institutions, purchased 3,700 DNA samples from the AGRE database to conduct the most comprehensive genetic analysis to date. AGRE Steering Committee member Dr. Rudy Tanzi, who is from the Massachusetts General Hospital and Harvard Medical School, is one of the leading investigators for this project.

Collaborations remain a key part of AGRE's mission and sharing our data with other researchers demonstrates the incredible opportunity to increase the pace of autism research. Our current collaborations include:

- AGRE and Stanford University continue to collect data for the California Autism Twin Study (CATS). To learn more details about this project read our article, *Largest California Population-Based Study of Autism Twins* on page 3.
- In August of this year, AGRE participated in four independent federal grant submissions under the Autism Centers of Excellence (ACE) program. The ACE program represents a consolidation of two existing federally funded programs, the Studies to Advance Autism Research and Treatment (STAART) and Collaborative Programs of Excellence in Autism (CPEA), in order to maximize coordination and cohesion of NIH-sponsored efforts in autism research.
- Cure Autism Now, AGRE, and the Southwest Autism Research & Resource Center (SARRC) will join forces to bring the first WALK NOW to Tempe Beach Park, AZ. Scheduled for November 19th, the proceeds from this family event will support continued data collection from families with two children affected by autism, families with one affected child, and a sample of families with typically developing children. To learn more about this event please visit: www.walknow.org
- Earlier this year AGRE, along with collaborators from Michigan State University and the Center for Autism and Developmental Disorders (CLIMA) in Mexico City, submitted a grant to the NIH to establish the infrastructure for autism research in Mexico. Dr. Carlos Marcin, Director of CLIMA, has brought together researchers, educators and clinicians in Mexico. In addition, the International Meeting for Autism Research, held earlier this summer in Montreal, brought together researchers from Spain, Venezuela, Nicaragua and Mexico to identify ways that they could work together to expand upon this grant proposal and replicate some of the research protocols currently being carried out in Spain and Latin America. All data collected in Mexico and Venezuela will be made available through AGRE.

Let me use this opportunity to express my deep gratitude to the AGRE families, to my hard-working staff, to Cure Autism Now, to the AGRE Steering Committee, and to the AGRE research community for allowing me to participate in such an outstanding crusade. Be assured, we will not stop until we reach our goals.

Thank you,
Clara Lajonchere

Since 2000, the AGRE clinical team has made 617 in-home clinical visits, 698 phlebotomy visits, and 308 medical visits, the majority conducted in the last 3 years. AGRE remains the single largest private collection of autism families in the world.

AGRE Update—Data Sharing Success

AGRE’s unique goal to share data with the scientific community remains strong, and we could not have done this without the overwhelming support and participation of our families. You are important partners in the effort to identify the causes and ultimately a cure for autism, and we thank you for everything that you do!

Recruitment—Hundreds of Families Want To Contribute

AGRE has had overwhelming success with family recruitment this year. Last year, we had 320 families sign up to be part of AGRE. To date, there are already close to 275 families across the nation that have signed up, and we are only in August!

Due to the tremendous jump in family recruitment, we are doing our best to see you and your family. Most of the AGRE staff members are located in our main office in Los Angeles, CA, and since they travel throughout the 50 states to see all of our families, there can be up to a year wait (or longer) for an in-home visit. If you have not heard from us in a while, please remember that we have not forgotten about you! We hope to be in your family’s area soon.

No matter which phase of the participation process you are in, it is important for us to have your family’s most up-to-date contact information. We may need to contact you to finish data collection, report research findings, or tell you about future collaborations. If you have changed your address, phone number or email, please contact us toll-free at 866-612-2473 or at info@familyagre.org.

Family Satisfaction

Families who have participated in AGRE are asked to complete a satisfaction survey designed to improve communication and better understand families’ needs. In 2006, responses from 74 families highlighted the overall positive view families have of AGRE, while pointing out that feedback is a priority for families, who want to know about research updates and important scientific findings.

Researcher Access

AGRE’s 2006 research activities include:

- Researchers can access information from over 800 families for use in their investigations.
- 160 AGRE-approved researchers from 13 countries have applied to study families in the AGRE database.
- AGRE samples were cited in more than 78 publications, 15 since the beginning of 2006, including major medical and science journals. Additionally, several AGRE abstracts were presented at professional conferences, including the 2006 International Meeting for Autism Research (IMFAR). The current list of papers citing the AGRE resource is now available on-line at: www.agre.org

Largest California Population-Based Study of Autism Twins

AGRE is working on the California Autism Twin Study (CATS) with Dr. Joachim Hallmayer at the Stanford University School of Medicine and collaborators at Kaiser Permanente, The California Department of Health and Human Services, and the MIND Institute at U.C. Davis.

The purpose of the study is to help researchers get a better understanding of the genetic basis of autism. Over a 5-year period, the CATS study hopes to visit 300 families with twin children where at least one child has autism. By studying twins, researchers also hope to learn whether some characteristics of autism can be tied to the twins’ shared pre- and post-natal environment. So far, AGRE has visited 29 families in Southern California, and the Stanford team has seen 10 families in Northern California, for a total of 39 twin pairs.

Twin studies are critical to understanding the heritability of any disorder. The CATS study will give researchers a better understanding of the interplay between genes and the environment—that is, which characteristics of autism are more influenced by genes, and which characteristics are more affected by the environment. Because the majority of these CATS twin families will be enrolled in the AGRE program, AGRE will house the largest collection of well-characterized twins with autism in the nation. ■

Key Contact Info

Recruitment

New families who want information about AGRE or have questions about enrollment contact:

Tiffany Torigoe, Family Recruitment Manager at
866-612-2473 x732

Conor McCann, Family Recruitment Specialist at
866-612-2473 x734

info@familyagre.org

Family Contact Information can be submitted online at:
www.autismtools.org/familyagre/register/

An AGRE brochure is available to download at:
www.cureautismnow.org/agre

Enrolled AGRE Families

Families who have questions regarding scheduling a visit to your home, or “next step” in participation contact:

Tiffany Torigoe, Family Recruitment Manager at
866-612-2473 x732

Conor McCann, Family Recruitment Specialist at
866-612-2473 x734

info@familyagre.org

Research Questions

Researchers and families who have research questions about AGRE contact:

Vlad Kustnovich, Researcher Liaison and
ISAAC Project Manager at 866-612-2473 x731

info@agre.org

Cure Autism Now

888-8 AUTISM
Fax 323-549-0547
info@cureautismnow.org
www.cureautismnow.org

Mail Address:

Cure Autism Now
AGRE
5455 Wilshire Blvd. Suite 2250
Los Angeles, CA 90036-4234



AGRE Funding

In 2006, Cure Autism Now will continue its partnership with researchers at the University of California, Los Angeles (UCLA). In addition, grant support from the National Institute of Mental Health will continue to further enhance the AGRE family collection.

Institutional Review Board (IRB)

AGRE is committed to ensuring the utmost protection and privacy of all our research participants. Any research study involving people must be overseen by an Institutional Review Board (IRB)—an outside agency that reviews and monitors biomedical research with human subjects. This agency reviews all consent forms, assessment materials, and recruitment documents to make sure that they comply with state and federal regulations involving human research. AGRE must update its IRB approval each year in order to continue its recruitment and data collection activities.

AGRE's primary IRB is the Western Institutional Review Board (WIRB), one of the oldest and most experienced independent IRB's in the United States. WIRB has provided review services for more than 10,000 investigators in over 30 countries and in all 50 United States. AGRE is also overseen by the IRB at the University of California, Los Angeles (UCLA), because of funding received through our UCLA collaboration with Dr. Daniel Geschwind and colleagues.

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

AGRE Collaborations

AGRE believes that collaborations with other groups and projects will provide the largest contribution to autism research. Therefore, we have been increasing our involvement with new collaborations to meet our goals. Below is a list of AGRE's collaborations:

- Washington University in St. Louis—Social Responsiveness Scale (SRS)
- UCLA—Brain Imaging Study
- Stanford University—California Autism Twin Study
- University of North Carolina—Language Study (Pending)

Co-Recruitment

- SARRC—Southwest Autism Research and Resource Exchange ■

Spotlight: Dr. Gary Stobbe

By Dr. Sarah Spence, M.D., Ph.D.



Dr. Stobbe obtained his Bachelor of Science degree in Psychobiology at UCLA in 1985 and Doctor of Medicine degree at Albany Medical College in New York in 1989. He then completed his neurology residency at UCLA Wadsworth VA Medical Center and Neuropsychiatric Institute where he first became interested in autism spectrum disorders.

In addition to his invaluable role as practicing physician and Medical Director at ASTAR (www.astarcenter.org), Dr. Stobbe visits AGRE families in their homes.

I first met Dr. Stobbe at an investigator's meeting and we immediately formed a connection because of our like-minded attitudes about pushing autism research forward and finding answers quickly. It was soon after that I was successful in recruiting him to work part-time in his "spare time" for AGRE. Since he began seeing families for us, I have heard nothing but rave reviews from everyone who has had the privilege of working with him—from families who invite him into their homes for the assessments to AGRE staff who adore his enthusiasm, easy-going nature and consummate professionalism.

Even more impressive is his continued quest for knowledge regarding autism spectrum disorders. Dr. Stobbe regularly attends scientific meetings that update him on state-of-the-art autism research. In a field where the research is changing so rapidly, it is imperative for providers to be up-to-date on the latest results. However, in real life practice, that is almost impossible to do. Amazingly, Dr. Stobbe finds a way to do it... and does it well.

Dr. Stobbe combines all the qualities that make a phenomenal provider for patients with autism and their families. He is smart, committed, compassionate, and most of all, driven by a passionate desire to truly understand this complex disorder. We are thrilled to have him on the AGRE team. ■

As part of AGRE's participation, an AGRE Physician visits families in their homes to collect family histories and look for traits and genetic syndromes that may predispose individuals and families to autism. Family histories are collected on all participating members; physical exams are completed on all participating siblings.



Jane Pickett visits Clara Lajonchere, AGRE Program Director

AGRE Families Urged to Learn About Autism Tissue Program

The Autism Tissue Program invites AGRE Families to find out more about how to register to become a brain tissue donor and make the important decision to participate in future brain tissue research.

AGRE Families can make an informed decision by learning more about the Autism Tissue program, and be prepared to act if an unexpected and unfortunate event were to occur.

The Autism Tissue Program can answer any questions you might have about how to register for a brain tissue donation. All inquiries are treated with absolute confidentiality.

Autism Tissue Program

99 Wall Street
Princeton, NJ 08540

Director: Jane Pickett, Ph.D.

Voice: 877-333-0999

Fax: 858-694-0748

Email: registry@memoriesofhope.org

Staff Announcements



AGRE Team and Dr. Spence

Sarah Spence, M.D., Ph.D., Medical Director, 2000–2006

Pursues New Career at NIMH

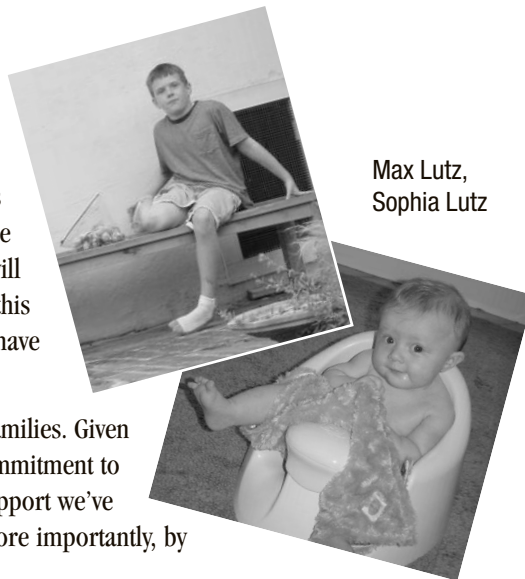
Dr. Spence has served as the AGRE Medical Director for the past six years. In June, Sarah transitioned to a new research career at the National Institute of Mental Health (NIMH). She is the Pediatric Neurologist on the new multidisciplinary autism research team in the Behavioral Pediatrics branch of the Institute. This team will be carrying out many exciting trials investigating the causes, clinical features and treatments of autism spectrum disorders. Sarah credits the AGRE families for her real training as an “autism expert”—in 2000 to 2001, Dr Spence visited over 100 families in their homes. She is eternally grateful to her teachers—the families who participate in the AGRE program.

Marin Lutz, Collaborations Manager, 2003–2006

Thank You for this Incredible Opportunity

After much thought and with a heavy heart, I have made the difficult decision to leave my position as AGRE’s Collaborations Manager. With the new addition of my daughter, it became clear that I needed to devote my time to her at home. One of the reasons I was drawn to AGRE is because my nephew Max is diagnosed with autism, and I wanted to be part of an organization that provided a collaborative approach to science. Although I will no longer be an official part of AGRE, I know I will always be a permanent part of this extended family. I have truly enjoyed this unique opportunity and feel fortunate to have worked with such a talented and dedicated team.

We couldn’t provide this unique resource without the tireless support from our AGRE families. Given the challenges and time constraints they face, I am overwhelmed by their continued commitment to this program and their willingness to participate in on-going collaborations. It is the rapport we’ve generated with these families that sets us apart. I am humbled by their support and, more importantly, by the level of devotion they provide their children.



Max Lutz,
Sophia Lutz

There is no place like it...Aspen, Colorado!

Brianne E. Cohen, Senior Clinical Research Associate
Autism Genetic Resource Exchange (AGRE)

This summer I had the opportunity to volunteer as a counselor at an amazing camp called *Extreme Sports Camp*, a sleepover camp for kids with autism in magnificent Aspen, Colorado. Between the surrounding beauty of Aspen and the fantastic kids, it turned out to be the highlight of my summer. Whether the activity was white water rafting, horseback riding, tubing or hiking, these children were immersed in a variety of outdoor activities from day until night.

The philosophy of *Extreme Sports Camp* is to provide the opportunity and the underlying instructional approach to experience the more advanced, age-appropriate physical activities that truly challenge children with autism. First handedly, I witnessed some amazing breakthroughs; a child who was afraid of riding horses (and wearing a helmet) conquered her fear through support and instruction from the staff. A child, who at first refused to step foot into the water raft, was later caught smiling as we rafted down the river.

It was such a joy to observe as well as report to the parents at the end of each day what their child had accomplished. It is a continuous learning experience for me to see the growth potential in these children. For more information on *Extreme Sports Camp*, please visit:

www.extremesportscamp.com



Danny Dubrowsky, age 11
and Brianne Cohen

We Can Make a Difference

By Dusan Bosnjakovic, ISAAC Project Coordinator

One day last spring, Dr. Clara Lajonchere, AGRE's Program Director, called me into her office to tell me about her recent trip to Chicago where she met a generous donor to Cure Autism Now (CAN) and also a father of an autistic boy. While dining in his home, Dr. Lajonchere learned about his dream of helping a school for special needs students in Bosnia and Herzegovina. He had visited the post-civil war region several times and was struck by the disproportionate population of children with special needs. Of special concern was the dire need for specialized educational services for these children. Because the area was so torn by years of very violent warfare, our CAN donor, who wanted to stay anonymous, felt that any positive action would have great impact.

Immediately our donor became immersed in the project. He formed "Team 1"—a team of experts to go to the region to do formal needs assessment and to begin the project design. The team consisted of doctors, speech therapists, physical therapists, and special education teachers. Because of my background of working in the field of autism and having lived half of my life in the former Yugoslavia, I became the seventh member of the team. My ability as an interpreter/translator would be useful to the team. Delighted with such a wonderful opportunity, I would soon be en route to Herzegovina, a land that held a special place in my heart.

In Siroki Brijeg, we visited Our Lady Hope, a local elementary school that serves thirty children with special needs. The twelve staff and parents welcomed us. We were impressed with the staff's dedication and level of caring as well as the involvement of the parents. We learned the school was not meeting the educational challenge of children with special needs. Our team committed to help them do this.

After giving the students some small gifts of toys and resources for their classrooms, we became busy working toward our time-limited goal. Each team of professionals paired up with their counterparts and spent several days with each child, assessing their weaknesses, planning appropriate steps toward rehabilitation, and demonstrating the educational approach and tools that work well in the schools back home. Serving as an interpreter, I had many opportunities to share information about autism with parents who had children af-



Team 1

ected by it. As the primary interpreter, I had the good fortune of being exposed to every facet of the mission. I had to comprehend and then focus on the broad curiosities of the staff and parents, as well as to distill the essence and relevance of our team's technical knowledge.

In war-torn countries such as most of the states of the former Yugoslavia, parents often do not have the strength, time, or education to understand the frustrations and vulnerability of their special needs child. So, many of these cases go unrecognized and untreated. Overworked doctors were blamed, the poorly functioning government was blamed and many parents even blamed themselves for the struggle their special needs children had to go through. At no time did we hear of or see blame directed at the children. I remember one mother who thought that because one of her children had undergone serious surgery and she devoted more time to him around the time that her other child was diagnosed with autism, that she had somehow caused his low-functioning autism. She had found misinformed supporting articles on the topic on the internet to further her belief that caused her to break down in front of me. One of the most satisfying moments that I experienced during this trip was when she heard and accepted our explanation of how she was not the cause of her son's autism.

The last day at the school, the team organized a meeting with all the parents, students, and staff. Following a brief introduction by our child psychiatrist, we began the Q & A section. The questions seemed to never end. The deep level of parental caring was so evident with each question. The testaments of what they had tried also demonstrated their best hopes for their children's future. Our team was quite moved by this. The several hour-long experience left everybody in the room inspired but rather tired.

The staff of Our Lady Hope treated "Team 1" to a farewell dinner at the best local restaurant where they expressed their gratitude for our help and we shared our appreciation for their hospitality and willingness to let us assist. We complimented the staff and parents further for their absolute dedication to their children.

Just before we left, our benefactor donated funds to the school for the construction of an additional school wing, an area that will be dedicated to the education of children with autism. We also left with a promise that there will be a "Team 2" to provide further help. I personally promised to join that phase of the project. ■



Our Lady Hope school
 Photo courtesy of Jesse Eisenhardt

AGRE Staff



Jocelyn Furr, Data Analyst, received her Bachelor's degree in Biology from Boston University. During her senior year in college, she interned for the Research and Development Department at a small biotech company in Boston that specialized in rare genetic disorders. After graduation, she was offered a job in the Quality Control Department at the same company, controlling the receipt and distribution of biological samples and data. She moved to Los Angeles in January 2004 and joined the AGRE team that February. She handles all analysis requests needed by upper management, collaborators, and grant reviewers. She is also responsible for managing the data that is collected by the clinical research associates and pediatric neurologists, as well as maintaining the quality of the internal and web-based databases and patient files. She is excited to devote her talents to help maintain the quality of data available to AGRE researchers, and is dedicated to AGRE's mission. Email her at jfurr@agre.org



Nancy Hart, AGRE Data Projects Manager, has been involved with Cure Autism Now (CAN) for the past eight years. Nancy first became involved with CAN as a volunteer, before she began working directly for AGRE in 2001, and currently manages the AGRE Data Projects team. In order to track the data for the families enrolled in AGRE, Nancy developed the System for AGRE Recruitment and Handling (SARAH), a database which houses and integrates the genetic, medical, and diagnostic information on families and produces the AGRE Pedigree Catalog. In addition, Nancy is the Program Manager for the Internet System for Assessing Autistic Children (ISAAC), which is quickly becoming a standard data management tool in the autism research community. Nancy has a 12-year-old son with autism and is committed to the missions of both CAN and AGRE. Email her at nhart@agre.org



Vlad Kustanovich, Ph.D., Researcher Liaison and ISAAC Project Manager, was born in Minsk, Belarus. He received a Bachelor of Arts in Molecular, Cellular and Developmental Biology and a Bachelor of Arts in Psychology from the University of California, Santa Cruz in 1996. Vlad has completed Doctoral studies in the field of human genetics and has received his Ph.D. from UCLA's new department of Human Genetics. He has recently expanded his role in AGRE to managing the operations of the ISAAC Database system, which stores and distributes to scientists anonymous information on study subjects in AGRE as well as in other studies. Email him at vlad@agre.org



Conor McCann, Family Recruitment Specialist, graduated from California State University, Northridge in 2004 with a Bachelor's degree in Philosophy. Conor started working for AGRE in April 2006 as the Research Assistant. He was recently promoted to the Family Recruitment Specialist position. Conor has been an excellent addition to the AGRE team. Email him at cmccann@agre.org



Janet Miller, J.D., Ph.D., Clinical Director, joined AGRE in August 2001. She received a B.A. in Psychology from U.C. Berkeley, a Ph.D. in Clinical Psychology from the Wright Institute, and a J.D. in Law from UCLA. Dr. Miller oversees the assessment and diagnosis of AGRE participants, trains and supervises the clinical team, and works extensively with AGRE research collaborations. She uses her legal training to negotiate contracts with researchers accessing the AGRE resource. Dr. Miller is also responsible for Institutional Review Board compliance to ensure the protection of AGRE research participants. Janet is delighted to be working with such a dedicated and talented group of AGRE staff members. Email her at jmiller@agre.org



Rachel Shefelbine, Clinical Research Associate, graduated from the University of California, San Diego with a Bachelor's Degree in Psychology. After working at the MIND Institute in Sacramento, Rachel joined the AGRE team in 2005.



Tiffany Torigoe, Family Recruitment Manager, graduated from the University of Hawaii at Manoa with a Bachelor's degree in Psychology. She moved to California from Hawaii in January 2004 and joined AGRE that February. She is currently responsible for recruitment and intake of all AGRE families. She is planning to attend graduate school in the future and hopes to open a clinic for children with developmental disabilities in her native Hawaii. She feels that AGRE is a perfect fit for her because of her desire to work in the scientific community and make a difference in children's lives. Email her at ttorigoe@agre.org

AGRE Staff



Kelly Alston, Collaborations Manager, graduated from UCLA with a Bachelor's degree in English. Before taking a position with AGRE, Kelly volunteered his time working on the LA Walk Now committee. Kelly will be a great asset to our collaborations team. Email him at kalston@agre.org



Charlotte DiStefano, Clinical Research Associate, graduated from Harvard University in 2006 with a Master of Education, Mind, Brain and Education Major. Charlotte moved from Boston to Los Angeles in July and joined AGRE in August to begin her training. We are very excited to have Charlotte be a part of the AGRE team. Email her at cdistefano@agre.org



Dusan Bosnjakovic, ISAAC Project Coordinator, graduated from UC San Diego with a Bachelor's Degree in Psychology. Dusan recently returned from a humanitarian mission to Bosnia and Herzegovina along with a medical team. The trip's goal was to assess and aid the special education centers of the developing region. In the office, Dusan coordinates the Internet System for Assessing Autistic Children (ISAAC) clinical data sharing projects. Email him at dbosnjakovic@agre.org



Angie Fedele, Clinical Operations Manager, graduated from Wesley College with a Bachelor's Degree in Psychology. Before working for AGRE, Angie spent three years implementing treatment plans with children on the autism spectrum. Angie needed a change, but wanted to continue to make a difference in the autism world. She felt passionate about what Cure Autism Now stood for and particularly about AGRE's mission. Angie joined AGRE in October, 2000 when the office was located in Philadelphia. Besides seeing families as a clinical rater, Angie provides supervision and training to the entire clinical team. In addition to her duties at AGRE, for the past three years Angie has been working on cognitive development with an autistic child in Los Angeles. "Over the past 5 1/2 years of working for AGRE, I have been fortunate to meet the most amazing families and individuals with autism. Every child has touched my heart and I take each family's story with me as I continue my work in the field of Autism." Email her at afedele@agre.org



Linda Parry-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 and has visited over 200 AGRE families.



Brianne E. Coben, Clinical Research Associate, graduated from the University of Miami with a double major in Music Therapy and Music Education. As a Board Certified Music Therapist, Brianne specialized in working with children with autism. Brianne has volunteered with the AGRE program since April 2002 and started working as a full time Clinical Research Associate in April 2003. Her primary responsibilities include conducting in-home observations, interviews and assessments of AGRE children. Email her at bcohen@agre.org





Corinne Currier, 12, and Cameron Currier, 11

Currier Family, Gardner, Kansas

I would really like to thank Dani Ocampo and Brianne Cohen for all they did for my kids! Corinne 12, and Cameron 11, were diagnosed with autism when they were 4 and 5. We had many struggles, but like every family we take everything day by day and set our goals.

Corinne will be starting Wheatridge Jr. High and is excited about becoming a teenager; she loves to draw and one day work for Hallmark! Cameron goes to Nike Intermediate School and will be in 6th grade. "Weatherman Cam" is what he is known as to his classmates. He loves weather and to help his social and speaking skills Cameron reads the weather report every day over the loud speaker. He has even been to a local TV station to see how the real weather reports are given.

Thank you to AGRE for all that you are doing to help those with autism and what you are doing for the future for many families. ■

Continued from page 1

denial for long; trusted friends gently encouraged us to seek a professional evaluation. A diagnosis by qualified medical professionals gave us the strength to replace denial with acceptance, and an added zeal to seek full recovery for our son.

This same strength helped us face diagnosis for Samuel a year and half later. Hindsight has been our cruelest teacher, but has educated us a great deal as well. If I could offer one piece of advice it would be, "time is of the essence. Do not waste a minute; *everything* you do for your children will make a huge difference in their future."

Educating ourselves as parents in a number of areas was the best way to advocate effectively for our family. Implementing intensive early intervention was critical. Building a solid community and a school-based support system for all of our children has been instrumental. We also believe healthy sibling relationships are crucial for the family as a whole.

Connecting to and participating with AGRE has been a wonderful opportunity for our entire family. We feel like we are contributing and making a difference in the autism research community. A cure is out there, recovery is possible, if we all work together. ■

Connor Jones is a 12-year-old boy with high functioning autism who has become an advocate at his school for other children with special needs and autism. Connor, his sister Sarah, and his mom Nancy live in Danville, California. Nancy has worked for CAN for the past eight years.

"As you see this paper I wrote, I will tell you about it. This was for the words of wisdom at my school for my teacher to read. It has to do with autism. The reason I wrote this is because my Core teacher had a words of wisdom (some pieces of advice a teacher reads every day) contest. The reason he had this contest is because the words of wisdom he was supposed to read was not that great. So a kid could write words of wisdom for extra credit. Two people wrote one. Someone else and I. I won. Mine was read aloud, but it remained anonymous of who it was. And here is the words of wisdom."

Connor Jones



Sarah and Connor Jones

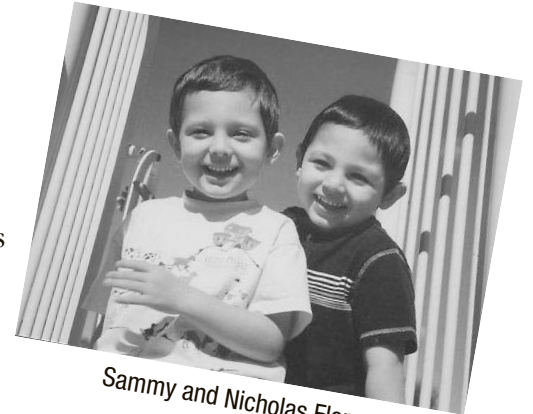
Words of Wisdom

Good morning! This is Leif Bostrom with your Words of Wisdom for March 1st, 2006, and a very special shout-out to all the philosophers in room 303. There are some people with special ed., including some kids at our school. But being special ed. does not mean that they are useless and will never do anything right. In fact, just last weekend, there was a boy with autism, in Rochester New York, who was manager on a basketball team. He was so enthusiastic about being manager, the coach gave him a shot in a jersey. In the last four minutes, he scored twenty points and sent mayhem not just across the bleachers, but the court as well. With something to think about, this is Leif Bostrom. Make it a great day, or not, the choice is yours.

“[autism]...It can be very devastating, but with the love and support of family and friends, I believe we can face just about anything.”

Flores Family, Grand Prairie, Texas

My husband Alan and I were truly blessed with our first child Nicholas in 1999. Nicholas was a normal child full of energy and hitting all his milestones. In February 2001, I gave birth to our second son Samuel and it was at that time we noticed that Nicholas had stopped talking. Over the next couple of months he seemed to withdraw into his own little world. He was still very loving and happy, but was in his own little world. In August 2002, he was diagnosed with autism just a few days short of his third birthday.

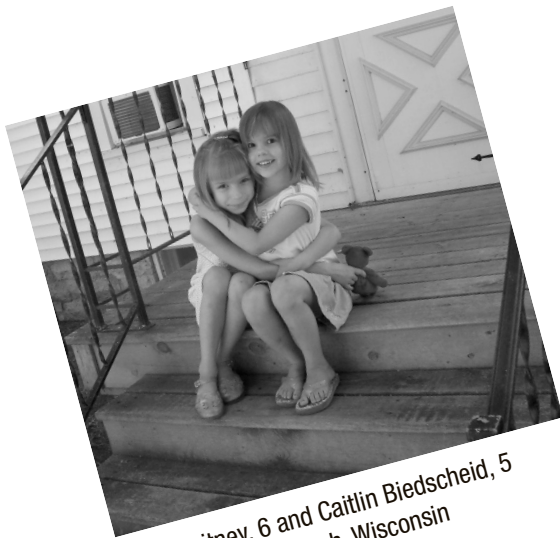


Sammy and Nicholas Flores

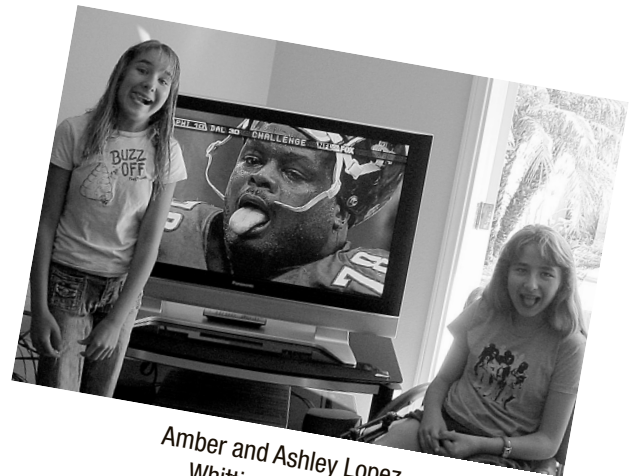
We watched Samuel very closely for any autistic traits; he babbled and hit all his first year milestones. We were told many times that autism could NOT affect both children. They were wrong. Samuel’s autistic traits turned out to be more severe—he had no verbal skills. He was diagnosed with autism around 26 months and we immediately started intensive intervention with him.

I ran across an article about AGRE in the CAN newsletter and immediately called about our family’s participation. The AGRE staff has been so helpful and caring about our family. It is very important to Alan and me to support researchers in finding a cure and ways to help children and families affected with autism. It can be very devastating, but with the love and support of family and friends, I believe we can face just about anything.

I am proud to say that Nicholas is regaining his language skills and is improving every day. Samuel started talking this last fall after completing the Tomatis Auditory Integration training. He continues to progress daily. The interaction of the boys together is wonderful. They play, wrestle and want to be together. They love airplanes and helicopters in the sky. They finally see the airplanes! ■



Britney, 6 and Caitlin Biedscheid, 5
Oshkosh, Wisconsin



Amber and Ashley Lopez
Whittier, California

We’re here for your kids, and now we want to hear from them!

We always love receiving photos and drawings from your kids—they brighten up our work space, and are a constant reminder of what we’re all working so hard to do. So we are asking for more—more photos, drawings, paintings, poems, stories, or anything else creative that your kids have made. Send them in, and we will hang them up around the office and even feature some in the next newsletter! We’re really excited, and hope your children will find this experience just as much fun!

Mail Address: AGRE
5455 Wilshire Blvd. Suite 2250, Los Angeles CA 90036



Difference between a “Research Evaluation” and a “Clinical Evaluation”

By Catherine Lord, Ph.D.

In general, the difference between a research evaluation and a clinical evaluation is the goal of the evaluation.

Research Evaluation

A **research evaluation** uses specific assessment tools that are reliable and have been validated by other researchers to answer a scientific question that researchers are studying.

In order to collect data for a research evaluation, families undergo an assessment that may be extremely comprehensive, sometimes seeming more thorough than a clinical evaluation. This reflects the wide range of data that is needed to answer complicated research questions that will be pursued using the data. Many, but not all, research projects provide reports so that families can review the results. Sometimes these reports can be used in clinical situations, particularly if the clinical team is planning to use the same assessment tools as the research team.

The research staff collecting data for a research evaluation may be very knowledgeable about autism. They are often graduate students or research associates with a keen interest in autism research and tend to be very informed about the field in general. They are not licensed clinicians, however, and do not have the professional training and working knowledge of diagnosis and treatment planning.

Clinical Evaluation

A **clinical evaluation** is an assessment of a particular child with an autism spectrum disorder that is designed to assist the family in making treatment choices. A licensed clinician, such as a clinical psychologist, psychiatrist or speech-language pathologist, completes the assessment.

A clinical evaluation should be organized specifically around your child’s needs or your family’s concerns. There may be a standard evaluation that is used in each case, but the focus of that evaluation should be on your child’s strengths, weaknesses, and the questions you have. The evaluation should be carried out (or at a minimum, supervised) by a licensed clinician who has experience in working with children/adults with autism. A clinical evaluation should result in a report. Typically, the clinician also provides an opportunity for the family to ask specific questions and learn about services that may be helpful to your child.

Participating in a research project can be a very positive experience, knowing that you are helping researchers better understand autism as well learning new information about your child’s development when you complete innovative research evaluations. Keep in mind, however, that research evaluations are not done for the purpose of educating you about your child or helping you to develop an intervention or educational plan—if that is what you need, it is important to have a clinical evaluation done as well. ■

Catherine Lord, Ph.D. is the Director of the University of Michigan Autism and Communication Disorders Center (UMACC), Professor of Psychology and Psychiatry, College of LSA and Senior Research Scientist, at the Center for Human Growth and Development (CHGD)

Researchers to Watch

Collins AL, Ma D, Whitehead PL, Martin ER, Wright HH, Abramson RK, Hussman JP, Haines JL, Cuccaro ML, Gilbert JR, Pericak-Vance MA.

Investigation of autism and GABA receptor subunit genes in multiple ethnic groups.

Neurogenetics. 2006 Jul;7(3):167–74.

A team led by Dr. Margaret Pericak-Vance at the Duke University Center for Human Genetics has extended previous findings of association between autism and GABRB1 and GABRB4 to African-American families with autism. These genes are receptors for a chemical called GABA which is important in reducing the levels of neural connections and chemical conversation in the nervous system. In addition, they have also identified that families with children that have both autism and seizures have a particularly significant association with genetic changes in GABRB1. Interestingly, Dr. Pericak-Vance and her colleagues were able to apply novel statistical analyses to show that there were interactions between the different genes such that autism was more associated with specific DNA changes at GABRB1 and GABRB4. These studies begin to address the multiple gene and multifactor analysis of autism. Families recruited into the **AGRE study** were used to supplement other families with autism recruited by Dr. Pericak-Vance and her lab.

Benayed R, Gharani N, Rossman I, Mancuso V, Lazar G, Kamdar S, Bruse SE, Tischfield S, Smith BJ, Zimmerman RA, DiCicco-Bloom E, Brzustowicz LM, and Millonig JH

Support for the Homeobox Transcription Factor Gene ENGRAILED 2 as an Autism Spectrum Disorder Susceptibility Locus

Am J Hum Genet. 2005 Nov 77(5):851–68.

Ryn Benayed, Neda Gharani and their colleagues tested EN2 as a candidate gene for autism spectrum disorders (ASD) because EN2 mouse mutants showed similar physical characteristics in the cerebellum to those reported for individuals with autism. In addition, the EN2 gene is located in a region on chromosome 7 that had provided suggestive evidence for linkage to ASD. In November of 2005, Benayed and colleagues replicated the finding of Gharani and colleagues (2004) of association between ASD and two genetic changes in the EN2 gene. Remarkably, the calculated risk due to the associated DNA changes may contribute to as many as 40% of ASD cases. Families recruited into the **Autism Genetic Resource Exchange (AGRE)** were used for the initial association study (Gharani et al., 2004) while a different set of AGRE families was used for the validation (Benayed et al., 2005).

Web Based Clinical Trial/Research Studies Listing

Recognizing the need for family information, CAN has developed a **Clinical Trial/Research Studies Web Listing** service. Central to this effort is providing information to help families with autism learn about the latest studies and make informed decisions about participating in autism research. This service lists new and on-going clinical trials/research studies throughout the world. Each listing provides key information about the study including a description of the study, criteria for participation, and direct contact information for the study's coordinator and principal investigators. Most importantly, we have made the research descriptions detailed enough so that you will understand the scientific rationale behind the study and be able to ask the coordinators informed questions.

We invite you to regularly visit www.cureautismnow.org/familyparticipation for updates and information about new research in your area.

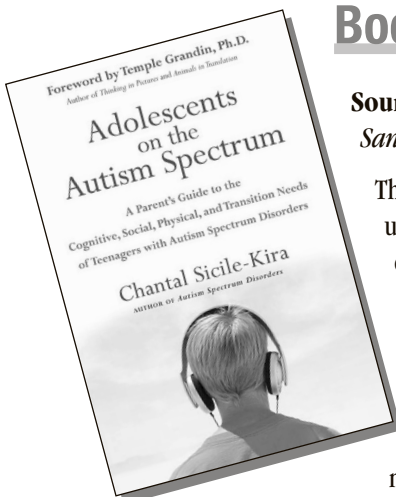
"I have been amazed at how passionate, caring, and well-informed some parents and families are who have been touched by autism. Being involved in research helps them to gain knowledge as well as a measure of control over what seems a random, devastating, and life-altering event. Their passion and energy motivates me, and others, to try to understand the causes and mechanisms of the disorder. Thus, the act of parental involvement itself attracts the resources to solve the problem and overcome autism."

Jaime A. Pineda, Ph.D.
Associate Professor
Department of Cognitive Science
University of California, San Diego

Research Facts

- In 2006 alone, over 300 researchers have applied for CAN grants.
- CAN-funded grants have resulted in 238 publications in major medical and science journals.

Bookstore



Sound Information About Adolescents and Autism

San Diego Family Magazine, April 2006

There's no doubt that raising a teen is challenging; raising a teen with an autism spectrum disorder offers unique challenges, and up until now, these challenges have not been illuminated in such a loving and honest way. *Adolescents on the Autism Spectrum: A Parent's Guide to the Cognitive, Social, Physical, and Transition Needs of Teenagers With Autism Spectrum Disorders* by local author Chantal Sicile-Kira is a shining example of a well-written parenting guidebook that offers practical and doable strategies to help teens navigate through puberty, hygiene issues, modesty and privacy issues, sexuality and social relationships. The author, the mother of an autistic teenager, draws from her own experiences with her son's autism and emergence into manhood: "The child on the autism spectrum who has difficulty with meltdowns and aggression may calm down at puberty. However, the teenage years are often a time when

tantrums appear and reappear. Usually these are due to frustration because like all teens, autistic teens want to have their

way and not always have to follow the rules that educators and parents set for them. Perhaps the teen is bored with being taught the same thing he didn't get in the special education class last year. Or perhaps the Asperger's teen doesn't understand the social cues and changes in his non-AS peers," Sicile-Kira writes. But there is hope: through education and communication, autistic teens can learn from their parents, teachers and medical professionals how to make a smoother transition into adulthood. ■

Chantal is the mother of a 17-year-old who is on the less able end of the spectrum with very limited speech. She deals frankly with adolescent issues that many people do not want to discuss such as sexuality. She deals with these issues in a practical, straightforward manner... Chantal's book also contains a great section on sibling and family issues.

RIDE NOW is a Rally, Ride, and Shindig

On September 17, 2006, RIDE NOW will be cruising through four cities (Chicago, Los Angeles, San Francisco, and Philadelphia/Atlantic City) to increase awareness for autism and raise money for research. Please visit www.canridenow.org for more information. Please email ridenow@cureautismnow.org to get more involved.

An Evening with the Stars

A Red Carpet Spectacular for Cure Autism Now

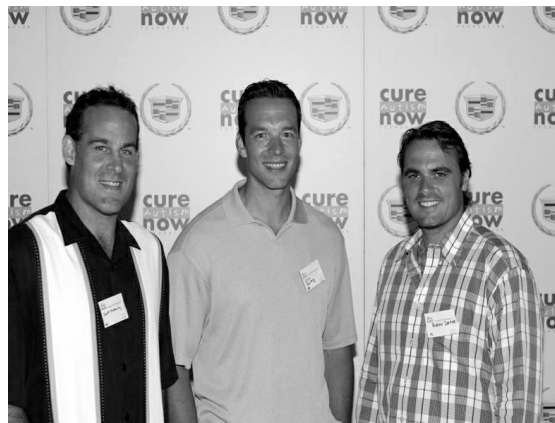
The Autism Community came together to celebrate Hollywood and raise \$40,000 for Autism Research at the Apollo Theater in Chicago in March of 2006. An Evening with the Stars is happening again! **Mark your calendars for Sunday, February 25, 2007! Please visit www.eveningwiththestars.org for more information!**

“Acts of Love: Dreams,” presented by Cadillac, will be held Sept. 18 at the Geffen Playhouse in Westwood, CA. Acts of Love 2006 will be a very special, one-night only celebration of dreams and dreamers performed by some of today’s finest actors, singers, and musicians. Please see www.cureautismnow.org/actsoflove for more information.

Fall Walk Season Preview

- 4th Annual **Philadelphia** WALK NOW on Saturday, September 30th at Citizens Bank Park
- 3rd Annual **Orange County** WALK NOW on Saturday, October 14th at Hidden Valley (Irvine, CA)
- 2nd Annual **Washington D.C.** WALK NOW on Saturday, October 21st on the National Mall
- 2nd Annual **New York/New Jersey** WALK NOW on Sunday, October 22nd at Battery Park in New York City
- 3rd Annual **Houston** WALK NOW on Saturday, October 28th at the Reliant Astrodome
- 3rd Annual **Seattle** WALK NOW on Saturday, September 30th at the University of Washington
- 2nd Annual **San Diego** WALK NOW on Saturday, November 4th at Balboa Park
- 2nd Annual **Orlando** WALK NOW on Saturday, November 18, 2006 at the Walt Disney World Wide World of Sports Complex
- 1st WALK NOW event in **Phoenix** — November 19th at Tempe Beach Park (This is a joint collaboration with SARRC)

WALK NOW is a true team effort, and we know that our volunteer leaders are the key to our success. Thank you for all your time, consideration, and effort to everyone who is contributing to the success of the Fall walks.



www.cureautismnow.org/chapters

Join US! Cure Autism Now chapters play a crucial role in helping Cure Autism Now raise funds for autism research and increase awareness of autism in local communities. Our chapters are made up of groups of fiercely determined parents, friends, and professionals who are not satisfied with the way things are today. They find strength in each other and are evidence of the fact that a group of committed people can indeed make a difference!

2006–2007 Chapters

Atlanta	Minneapolis
Chicago	New England
Cleveland	New York/New Jersey
Dallas/Fort Worth	Orange County
Denver	Orlando
Detroit	Philadelphia
Hawaii	San Diego
Houston	San Francisco Bay Area
Las Vegas	Seattle
Los Angeles	Washington, D.C.

www.cureautismnow.org

Are you connected? Sign up for Connections, Cure Autism Now’s eNewsletter and join the Cure Autism Now mailing list—receive news and updates to stay informed.

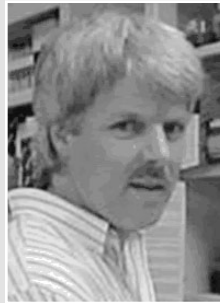


Continued from page 1

Over the past five years, scientists in the biological and medical sciences have developed amazing tools to identify general biological mechanisms and pathways involved in disorders such as cancer and heart disease, which are now being applied to the study of autism. Most prominent among these tools is the microarray or DNA chip. DNA chips are microscope slides with tens to hundreds of thousands of unique DNA spots on them. Each spot contains a unique set of DNA which can act as a target to “capture” a fluorescently labeled RNA that matches its sequence. Laser scanners are used to get a count of the amount of light coming off of each spot and the level of light corresponds to the amount of RNA captured by the particular spot. With tens to hundreds of thousands of spots, every single gene in the human genome is typically represented on a single DNA chip and the level and activity of every gene can be seen in one experiment.

A team led by Jeff Gregg, M.D. at the University of California, Davis used cell lines from a small number of **AGRE** participants to compare RNA levels in cells from participants with autism and their unaffected brothers or sisters. This was done in order to see which RNA's appear to be elevated or reduced in cells derived from autistic children in comparison to their non-autistic siblings. Interestingly, the researchers identified 48 RNA's which differed between the autistic and non-autistic individuals. These RNA's had a variety of important functions including cell development, cell signaling, metabolism and the response to external stimuli, indicating that the immune system may also be involved.

Valerie Hu, Ph.D. and her colleagues used similar tools to investigate if there were RNA's which differed in abundance between identical twins that had autism and those with a less severe autism spectrum disorder. Interestingly, RNA's related to nervous system development were found to be the most



*“The resources provided by **AGRE** have allowed me to examine important questions related to the etiology of Autism Spectrum Disorders.”*

Stephen J. Walker, Ph.D.
 Assistant Professor
 Wake Forest University School of Medicine
 Department of Physiology & Pharmacology

different between the two groups. In addition, RNA's related to inflammation and the immune response were also found to differ between cells from twins with the full autism diagnosis according to the Autism Diagnostic Interview and the cells from their siblings with another autism spectrum classification such as PDD-NOS. Finally, Dr. Hu and colleagues found that many of the RNA's which differed between the two groups were coded by genes in the vicinity of previously identified genetic association or in areas showing strong genetic linkage from purely genetic studies. This supports the previous genetic findings and those identified by these truly new and innovative experiments. These types of experiments may help shed light on not only the most severe aspects of autism, but may also help differentiate between the less severe subtypes of autism.

Finally, Drs. Walker, Segal and Aschner used cells from **AGRE** participants to investigate a very controversial topic, thimerosal. Thimerosal is an ethyl mercury based preservative previously found in many vaccine preparations such as those for hepatitis B,

diphtheria, pertussis, and tetanus (DPT) and measles, mumps and rubella (MMR). It has been suggested that autism may be triggered by thimerosal in individuals with a genetic predisposition. The research team proposed to investigate whether cell lines from children with autism differed in their levels of RNA's when treated with thimerosal from siblings who did not have autism. The researchers were especially interested in the changes in the RNA levels of metallothioneins, a set of proteins responsible for eliminating heavy metals from the cells. They tested the RNA levels in cells from both autistic and unaffected children, and then tested them in response to both ethyl mercury and zinc. Although the research team did not find significant differences between the response of autistic and non-autistic siblings to thimerosal, they did find that cell lines treated with ethyl mercury or zinc showed substantial increases in the levels of several RNA's. Treatment with zinc resulted in significant increases in the levels of metallothioneins. Some interesting RNA's that were found to be elevated in cells treated with ethyl mercury were the stress proteins. These RNA's are responsible for the activities of cells in stressful situations such as extreme heat, starvation, deprivation of water or oxygen and chemical toxicity (including metal toxicity). While these experiments show that ethyl mercury and zinc result in large changes in these cells, there is no evidence that there are any differences in the response of cells from autistic versus non-autistic study participants. This suggests that in this experimental model, cells from autistic individuals do not metabolize ethyl mercury in a significantly different way than their unaffected siblings. ■



The Cure Autism Now Foundation (CAN) was founded in 1995 with the goal of identifying the causes of autism and rapidly finding ways to prevent, treat and cure it through funding autism research, developing scientific resources, and raising awareness for the disorder that now affects one in every 166 children.

OUR CORE BELIEFS

Urgency Matters

- Autism is a national emergency
- A cure for autism will come during our children's lifetime

Improved Quality of Life is Possible

- Individuals with autism deserve a better quality of life
- Autism can be treated
- Early identification leads to better outcomes

Science is Key to the Solution

- A cure for autism is within reach
- The people who will solve autism are alive today
- Science can be hurried

OUR CORE VALUES

- Entrepreneurial, innovative and creative
- Aggressive and optimistic
- Possess a sense of urgency to find treatments and a cure
- Promote scientific excellence
- Promote scientific collaboration and broad data-sharing
- Recognize the value of a strong political voice and activism
- Leverage the passion of families

For more information, or to be added to our mailing list, please contact Cure Autism Now at (888) 8-AUTISM or info@cureautismnow.org.

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