



## CHILDHOOD AUTISM RISKS FROM GENETICS AND THE ENVIRONMENT

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Update April 2010

The goal of the CHARGE study is to understand possible ways that genetics combined with environment can influence a developing brain, leading to autism and other disabilities. Researchers have learned that autism is complex. Instead of looking for a single cause, we now understand that many factors probably affect how the brain develops. To study the combined effect of environmental exposure and genetics requires a large number of families. Thankfully, so many have chosen to participate in the CHARGE effort, we can now begin the task of analyzing the information collected from all of the families during their participation.

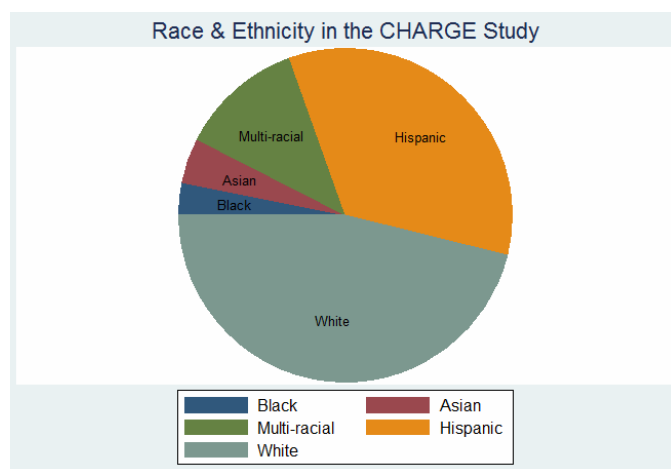
### Who is in the *CHARGE* study?



CHARGE study enrollment got underway in 2003. Since that time, nearly 1,400 families have participated, providing researchers with information that has led to several important scientific discoveries (see Science Update below). Of the families who have enrolled in the CHARGE study, 50% are families who have a child with autism, 22% have a child with developmental delays

without autism, and 28% have children that are developing typically.

Like California, CHARGE study families represent diverse ethnic and racial groups. About 32% of children in the CHARGE study are Hispanic or Latino, about the same percentage of Hispanic or Latino Californians in the 2000 U.S. Census. Percentages of Asian and African American children in the CHARGE study are somewhat lower than they are in California as a whole, reflecting the demographics of the population in the study's location. Geographically, families come from large cities, suburbs and rural communities. About 51% of children in the study were born in counties with a current population of a million or more people, while about 5% were born in counties with less than 100,000 people.



### What's new in *CHARGE*?

Last summer, the CHARGE study was awarded additional funds through the American Recovery and Reinvestment Act, which provided new research funding to our sponsor, the National Institutes of Health. With

this new funding, the CHARGE study is able to collect home dust samples to measure levels of pesticides, chemical flame-retardants (PBDEs), and chemicals that are added to plastic (Bisphenol-A and phthalates). Because some of these compounds stick to very fine dust particles that don't get picked up by regular household vacuum cleaners, the compounds can remain in the carpet for several years. With a special type of vacuum cleaner called the HVS3, we can collect these very fine dust particles and learn about early life exposures and their possible role in the development of autism. We are enrolling about 350 families for the dust study, which involves CHARGE study staff making an appointment to visit the home to collect the sample. To be eligible, families need to be living in the same home as when the enrolled child was born, or to have an area rug that was in the home when the child was born. To date, 60 CHARGE families have participated in this new study.

The CHARGE study staff and research team are committed to ensuring the best possible experience for families who decide to participate in the CHARGE study. That is why we consulted with parents on our Community Advisory Council when developing our clinic process back in 2001 before study enrollment began. Last summer, we developed a survey to provide parents in the study with an opportunity to give us feedback and suggestions, which is being used to evaluate and continually improve our process. These confidential surveys are mailed out along with the letter containing results from children's clinical evaluations. A postage-paid and pre-addressed envelope is included. So far, the feedback has been overwhelmingly positive. We appreciate the time many parents take to write detailed suggestions and comments. Based on suggestions we have received so far, we are working on finding ways to shorten the telephone interview and otherwise enhancing the participation experience for parents and children. CHARGE study parents who did not receive a survey

and would like one, please contact Melissa Rose, CHARGE study Project Manager at (916) 703-0219, or email [mbrose@ucdavis.edu](mailto:mbrose@ucdavis.edu).



### **Outreach and Education Update**

On January 11, CHARGE study staff member Crystal Gloria presented a workshop for health workers at La Clinica de La Raza in Oakland. The workshop was presented in Spanish and eleven people participated in the workshop, which provided information and resources for protecting pregnant women, infants and children from environmental hazards. The workshop was facilitated by Leticia Gonzales, Health Educator for LCDLR, and Teresa Ortiz, Project Manager for the National Children's Study, Sacramento site. The workshop was made possible by the Partnerships in Environmental Public Health (PEPH), a new program funded by the National Institute of Environmental Health Sciences.

### **Science Update**

To date, some of the most important findings from the CHARGE study have come from laboratory studies of the immune system, adding to a growing body of scientific evidence supporting a close relationship between the immune system and the developing brain. Cytokines are proteins necessary for communication between cells. Transforming Growth Factor Beta-1 (TGF- $\beta$ 1) is a cytokine involved in suppressing cells that cause inflammation. CHARGE study researcher, Paul Ashwood and colleagues found lower levels of TGF- $\beta$ 1 in children with ASD compared to children without an

ASD. They also found that lower levels of this cytokine were associated with more severe behavioral symptoms in children with ASD. The study, “**Decreased transforming growth factor beta1 in children with autism: A potential link between immune dysregulation and impairment in clinical behavioral outcomes**”, was published in *Journal of Neuroimmunology*.

Natural Killer cells (NK cells) play an important part in the immune system by providing immediate defense against infection from viruses. By studying children’s NK cells and the genes that regulate how they function, CHARGE study scientists in the laboratory of Dr. Frank Sharp, found differences that suggest children with an autism spectrum disorder, compared to typically developing children, may have abnormal NK cell function that could make them vulnerable to problems during brain development. The study titled, “**Altered gene expression and function of peripheral blood natural killer cells in children with autism**”, was published in the journal, *Brain, Behavior and Immunity*.

Some studies have suggested that there may be differences in mercury levels in children with autism compared to children without autism. In a recent paper, Dr. Irva Hertz-Picciotto reported measurements of mercury in CHARGE study children. In this study, children with autism or other ASD had similar blood mercury levels to children who are typically developing. Moreover, both groups had levels close to the mercury levels measured in a nationally representative sample of children in a similar age range. As expected, children who ate fish had higher levels than children who did not eat fish.

Because these mercury measurements were taken after children with autism had been diagnosed, the results cannot address the question of whether mercury plays a causal role in autism. The report, titled **Blood Mercury Concentrations in CHARGE Study**

**Children with and without Autism**, was published in the journal *Environmental Health Perspectives*.

None of these important scientific discoveries would have been possible without the children and families who have generously donated to the CHARGE study effort. For more information about the CHARGE study, please visit our website at <http://beincharge.ucdavis.edu/> or to request a hard copy of any CHARGE publication, please contact Melissa Rose at (916) 703-0219.

The CHARGE Study is sponsored by the National Institute of Environmental Health Sciences, The U.S. Environmental Protection Agency, with additional funding from Autism Speaks and the UC Davis M.I.N.D. Institute.



Photo: CHARGE Staff at Bay Area Walk Now for Autism

### **Acknowledgements**

The CHARGE Study is funded by the National Institute of Environmental Health Sciences (2P01ES001269 and R01ES015359), the U.S. Environmental Protection Agency (R833292 and R829388), Autism Speaks, and the UC Davis MIND Institute. We thank all of the CHARGE study children and their family members for their participation in this study.