



## PRESS ROOM

### News Releases

### About Children's

### News Clips

### New Research Building

### Public Affairs Staff/Policy

### Dream Magazine

### Pediatric Views

### Childrens News

#### FOR IMMEDIATE RELEASE

#### FOR IMMEDIATE RELEASE

Wednesday, Dec. 8, 2004

#### For Further Information:

Aaron Patnode

617-355-6420

<http://www.childrenshospital.org/cfapps/>

#### Children's Hospital Boston Launches Major Genetic Study of Autism

*Genetics, Genomics, Bioinformatics and Neuroscience Join Forces*

Children's Hospital Boston has begun enrolling patients as part of an ambitious new multidisciplinary study of autism that will attempt to pin down its genetic and biochemical causes. Results could be available in a year or two, and could yield a greater biological understanding of autistic spectrum disorders, better diagnostic and prognostic techniques, and potential medical treatments.

More than 90 percent of autism cases are believed to have a genetic component, and multiple genes are believed to be involved. But although much research has been done and many candidate genes identified, none have been clearly implicated. The Children's study, partially funded by the Nancy Lurie Marks Family Foundation, will bring together researchers in genetics, genomics, bioinformatics, and developmental medicine to try to solve the puzzle.

Autism is a complex behavioral syndrome defined by developmental deficits, particularly communication deficits, impaired social interaction and repetitive behaviors. Affecting about one in 1,000 people, it is now thought of as a spectrum of disorders including autism, Asperger's syndrome and pervasive developmental disorder. There is no specific medical treatment, although behavioral interventions help children on the autistic spectrum live fuller, more functional lives.

The Children's researchers plan to enroll 100 to 150 children age 2 years and older per year, along with their parents and affected siblings. They also will enroll 150 unaffected children to serve as controls. The study has several components:

✎ The Children's Developmental Medicine Center will first conduct detailed behavioral evaluations of the children and their families, led by Drs. Janice Ware and Leonard Rappaport. Subjects will be assessed for autistic spectrum disorders and carefully classified according to rigorous clinical research criteria. The goal is to develop behavioral profiles that can be correlated with genetic data. Children and their parents will then give saliva samples for DNA analysis and blood samples for RNA gene expression studies.

✎ Led by Drs. Ingrid Holm and Louis Kunkel, researchers in the Children's Program in Genomics will study the DNA samples, performing association studies and linkage studies to look for genetic differences (polymorphisms) that are shared within families and may accompany clinical manifestations of autistic spectrum disorders.

✎ The Program in Genomics also will perform microarray ("gene-chip") studies of RNA from white blood cells to examine differences in gene activity, or expression, among autistic children, their parents, and matched control subjects. By looking at 60,000 genes simultaneously and determining which are turned "on" and "off," the researchers will seek patterns or genetic "signatures" that mark the different autistic spectrum disorders and give clues to their biological causes. The investigators hope to show that gene expression in white blood cells is similar enough to that in brain cells to be a useful surrogate measure, avoiding the need to obtain and test brain tissue.

✎ The Children's Hospital Informatics Program (CHIP), led by Dr. Isaac Kohane, will perform computational analyses to help detect subtle genetic patterns, group patients according to their gene-expression profiles, and apply statistical techniques to determine the reliability of the patterns and linkages found and eliminate false-positive findings. As new data become available, CHIP will factor them in to create the strongest possible predictive models for autistic spectrum disorders.

✎ The Neurobiology Program of Children's Hospital, led by Dr. Michael Greenberg, will examine the connection between autism, a protein called brain-derived neurotrophic factor (BDNF), and several genes known to regulate BDNF. BDNF regulates many aspects of brain development and function, including formation of synapses. Mutation of one gene that regulates BDNF, called MeCP2, has already been linked to Rett syndrome, which is characterized by mental retardation and autistic behaviors. Further studies of MeCP2 and related genes will explore how they work and how brain-cell activity triggers them. These investigations may uncover other genes and biochemical pathways that underlie autism.

✎ Children and families enrolled in the Children's study also will be invited to take part in research at MIT's Department of Brain and Cognitive Sciences. Study participants will be tested for their ability to recognize and respond to faces (a critical deficit in autism), the development of specific language skills (also impaired in autism), and gross and fine motor function. Although not a formal part of the study, the data collected by MIT will be fed back into the Children's study and correlated with the genetic data.

Families can receive additional information on the autism study by contacting Lindsay Jackson at the Developmental Medicine Center (617-355-3076 or <http://www.childrenshospital.org/cfapps/>).

*Children's Hospital Boston is the nation's leading pediatric medical center, the largest provider of health care to Massachusetts' children, and the primary pediatric teaching hospital of Harvard Medical School. Children's provides pediatric and adolescent health services for patients from birth through age 21. In addition to 325 inpatient beds and comprehensive outpatient programs, it houses the world's largest research enterprise based at a pediatric medical center. More than 500 scientists, including eight members of the National Academy of Sciences, nine members of the Institute of Medicine and 10 members of the Howard Hughes Medical Institute comprise Children's research community. For more information about the hospital visit: <http://www.childrenshospital.org/cfapps/>.*

- ### -

Questions regarding the Press Room should be sent to  
[pubaff@tch.harvard.edu](mailto:pubaff@tch.harvard.edu)

---

[Children's Hospital Boston](#)  
300 Longwood Avenue  
Boston, MA 02115  
617-355-6420

Copyright © 2001, Children's Hospital Boston. All rights reserved.

The information on this website should not be taken as medical advice, which can only be given to you by your personal health care professional.

Updated 02/11/2005