

### **Recent Autism Science**

Thomas Insel, M.D.
Chair, IACC
IACC Full Committee Meeting
December 14, 2010

These slides do not reflect decisions of the IACC and are for discussion purposes only.



### Autism "Hot Topic" at SFN



In 2009: 255 abstracts
In 2010: 337 abstracts
and a public symposium



# A Model for Neural Development Cell 11/10 and Treatment of Rett Syndrome Using Human Induced Pluripotent Stem Cells

Maria C.N. Marchetto, 1,5 Cassiano Carromeu, 2,5 Allan Acab, 2 Diana Yu, 1 Gene W. Yeo, 3 Yangling Mu, 1 Gong Chen, 4 Fred H. Gage, 1 and Alysson R. Muotri 2,4

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<sup>2</sup>University of California San Diego, School of Medicine, Department of Pediatrics, Rady Children's Hospital San Diego, Department of Cellular and Molecular Medicine, Stem Cell Program, 9500 Gilman Drive, La Jolla, CA 92093, USA

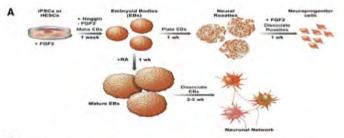
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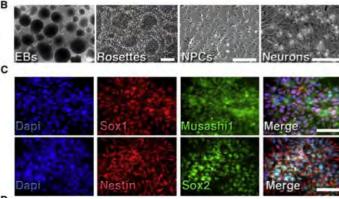
<sup>5</sup>These authors contributed equally to the work

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Rescue with IGF1 and Gentamycin



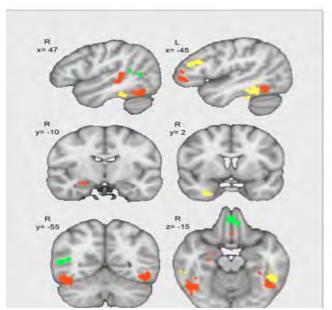


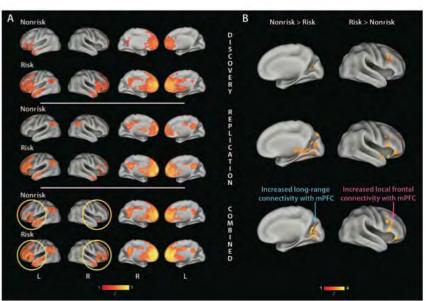
### Neural signatures of autism

PNAS 12/7/10

Martha D. Kaiser<sup>a</sup>, Caitlin M. Hudac<sup>a</sup>, Sarah Shultz<sup>a,b</sup>, Su Mei Lee<sup>a,b</sup>, Celeste Cheung<sup>a</sup>, Allison M. Berken<sup>a</sup>, Ben Deen<sup>a</sup>, Naomi B. Pitskel<sup>a</sup>, Daniel R. Sugrue<sup>a</sup>, Avery C. Voos<sup>a</sup>, Celine A. Saulnier<sup>a</sup>, Pamela Ventola<sup>a</sup>, Julie M. Wolf<sup>a</sup>, Ami Klin<sup>a</sup>, Brent C. Vander Wyk<sup>a</sup>, and Kevin A. Pelphrey<sup>a,b,1</sup>

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## Altered Functional Connectivity in Frontal Lobe Circuits Is Associated with Variation in the Autism Risk Gene CNTNAP2

Ashley A. Scott-Van Zeeland, 1,2,3 Brett S. Abrahams, 4\* Ana I. Alvarez-Retuerto, 4,5
Lisa I. Sonnenblick, 4 Jeffrey D. Rudie, 2 Dara Ghahremani, 6 Jeanette A. Mumford, 7
Russell A. Poldrack, 7 Mirella Dapretto, 5,6,8 Daniel H. Geschwind, 4,5+# Susan Y. Bookheimer 1,5,6,7+#

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### Describing the Brain in Autism in Five Dimensions—Magnetic Resonance Imaging-Assisted Diagnosis of Autism Spectrum Disorder Using a Multiparameter Classification Approach

Christine Ecker,<sup>1</sup> Andre Marquand,<sup>2</sup> Janaina Mourão-Miranda,<sup>3,4</sup> Patrick Johnston,<sup>1</sup> Eileen M. Daly,<sup>1</sup> Michael J. Brammer,<sup>2</sup> Stefanos Maltezos,<sup>1</sup> Clodagh M. Murphy,<sup>1</sup> Dene Robertson,<sup>1</sup> Steven C. Williams,<sup>3</sup> and Declan G. M. Murphy<sup>1</sup>

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The Journal of Neuroscience, November 3, 2010 - 30(44):14595-14609 - 14595

Neurobiology of Disease

Changes in Prefrontal Axons May Disrupt the Network in Autism

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### Mitochondrial Dysfunction in Autism

Cecilia Giulivi, PhD	
Yi-Fan Zhang, BS	
Alicja Omanska-Klusek, MS	
Catherine Ross-Inta, BS	
Sarah Wong, BS	
Irva Hertz-Picciotto, PhD	
Flora Tassone, PhD	
Isaac N. Pessah, PhD	

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UTISM SPECTRUM DISORDERS

**Context** Impaired mitochondrial function may influence processes highly dependent on energy, such as neurodevelopment, and contribute to autism. No studies have evaluated mitochondrial dysfunction and mitochondrial DNA (mtDNA) abnormalities in a well-defined population of children with autism.

Objective To evaluate mitochondrial defects in children with autism.

**Design, Setting, and Patients** Observational study using data collected from patients aged 2 to 5 years who were a subset of children participating in the Childhood Autism Risk From Genes and Environment study in California, which is a population-based, case-control investigation with confirmed autism cases and age-matched, genetically unrelated, typically developing controls, that was launched in 2003 and is still ongoing. Mitochondrial dysfunction and mtDNA abnormalities were evaluated in lymphocytes from 10 children with autism and 10 controls.

#### Parental Autoimmune Diseases Associated With Autism Spectrum Disorders in Offspring Epidemiology 9/10

Alexander Keil, Julie L. Daniels, Julia Forssen, Christina Hultman, Sven Cnattingius, Karin C. Söderberg, Maria Feychting, and Par Sparen



PNAS 6/10

## Characterization and reversal of synaptic defects in the amygdala in a mouse model of fragile X syndrome

Aparna Suvrathana, Charles A. Hoefferb, Helen Wongb, Eric Klannb, and Sumantra Chattarjia,1

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