

# Scientific Update

**Thomas Insel, M.D.**

Director, National Institute of Mental Health and Chair, IACC  
IACC Full Committee Meeting – July 19, 2011

# When should I be concerned?

THE JOURNAL OF PEDIATRICS • www.jpeds.com

ORIGINAL  
ARTICLES

## Detecting, Studying, and Treating Autism Early: The One-Year Well-Baby Check-Up Approach

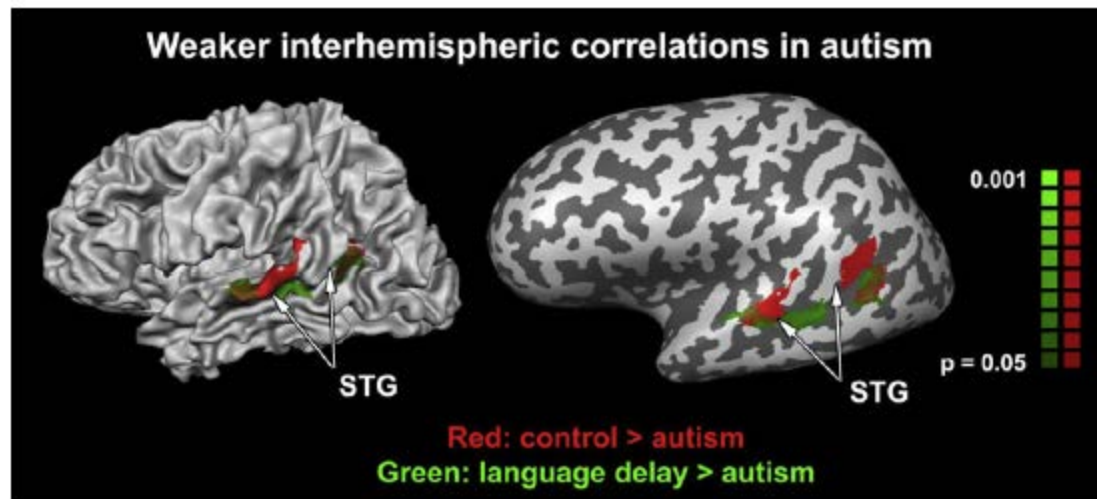
Karen Pierce, PhD, Cindy Carter, PhD, Melanie Weinfeld, PhD, Jamie Desmond, MPH, Roxana Hazin, BS, Robert Bjork, MD, and Nicole Gallagher, BA

## Disrupted Neural Synchronization in Toddlers with Autism

Neuron  
Report

Ilan Dinstein,<sup>1,4,\*</sup> Karen Pierce,<sup>1,3</sup> Lisa Eyer,<sup>1,2</sup> Stephanie Solso,<sup>1</sup> Rafael Malach,<sup>4</sup> Marlene Behrmann,<sup>5</sup> and Eric Courchesne<sup>1,3</sup>

<sup>1</sup>Autism Center of Excellence



# How can I understand what is happening?

## How common is autism?

CATHERINE LORD

**nature** June 9, 2011

RESEARCH

NEWS & VIEWS

AJP in Advance. Published May 9, 2011 (doi: 10.1176/appi.ajp.2011.10101532)

Article

## Prevalence of Autism Spectrum Disorders in a Total Population Sample

Kim YS, Leventhal BL, Koh YJ, Fombonne E, Laska E, Lim EC, Cheon KA, Kim SJ, Kim YK, Lee H, Song DH, Grinker RR

TABLE 3. Prevalence Estimates of Autism Spectrum Disorders (ASDs) in a South Korean Community<sup>a</sup>

Measure	Prevalence (%)	95% CI
Population <sup>b</sup>		
Total population	2.64	1.91–3.37
General-population sample	1.89	1.43–2.36
High-probability group	0.75	0.58–0.93
ASD Type		
Any ASD	2.64	1.91–3.37
Autistic disorder	0.94	0.56–1.34
Other ASDs <sup>c</sup>	1.70	1.08–2.32
Sex		
Male	3.74	2.57–4.90
Female	1.47	0.60–2.37

Total ASD = 2.64  
Autistic Dis = 0.94

<sup>a</sup> The denominator used in computing prevalences was all 55,266 children 7–12| years old in the study population. Statistical adjustments were made for nonparticipants and missing data.

# How can I understand what is happening?

ARCH GEN PSYCHIATRY/VOL 68 (NO. 5), MAY 2011

WWW.ARCHGENPSYCHIATRY.COM

459

## Epidemiology of Autism Spectrum Disorders in Adults in the Community in England

*Traolach S. Brugha, MD(NUI), FRCPsych; Sally McManus, MSc; John Bankart, MSc, PhD; Fiona Scott, PhD, CPsychol; Susan Purdon, MSc, PhD; Jane Smith, BSc; Paul Bebbington, PhD, FRCPsych; Rachel Jenkins, MD, FRCPsych; Howard Meltzer, PhD*

## PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

## Trends in the Prevalence of Developmental Disabilities in US Children, 1997–2008

*Coleen A. Boyle, Sheree Boulet, Laura A. Schieve, Robin A. Cohen, Stephen J. Blumberg, Marshalyn Yeargin-Allsopp, Susanna Visser and Michael D. Kogan*

<u>Dx</u>	<u>n (unwtd)</u>	<u>% all yrs</u>	<u>% change</u>
Any DD	15956	13.87	17.1%
ADHD	7652	6.69	33.0%
Autism	537	0.47	289.5%
Intell Dis	868	0.71	-1.5%

# How can I understand what is happening?

nature

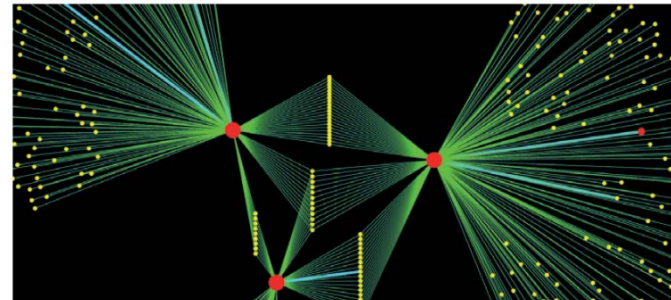
DISEASE GENETICS

## Converging models for autism

NATURE | VOL 474 | 16 JUNE 2011

### Transcriptomic analysis of autistic brain reveals convergent molecular pathology

Irina Voineagu<sup>1</sup>, Xinchun Wang<sup>2</sup>, Patrick Johnston<sup>3</sup>, Jennifer K. Lowe<sup>1</sup>, Yuan Tian<sup>1</sup>, Steve Horvath<sup>4</sup>, Jonathan Mill<sup>3</sup>, Rita M. Cantor<sup>4</sup>, Benjamin J. Blencowe<sup>2</sup> & Daniel H. Geschwind<sup>1,4</sup>



### Protein Interactome Reveals Converging Molecular Pathways Among Autism Disorders

Yasunari Sakai,<sup>1,2</sup> Chad A. Shaw,<sup>2\*</sup> Brian C. Dawson,<sup>1,2</sup> Diana V. Dugas,<sup>2</sup> Zaina Al-Mohtaseb,<sup>2</sup> David E. Hill,<sup>3</sup> Huda Y. Zoghbi<sup>1,2,4,5,6\*</sup>

8 June 2011 Vol 3 Issue 86 86ra49

# What caused this to happen and can it be prevented?

ARCH GEN PSYCHIATRY PUBLISHED ONLINE JULY 4, 2011 WWW.ARCHGENPSYCHIATRY.COM

## Genetic Heritability and Shared Environmental Factors Among Twin Pairs With Autism

*Joachim Hallmayer, MD; Sue Cleveland, BS; Andrea Torres, MA; Jennifer Phillips, PhD; Brianne Cohen, BA; Tiffany Torigoe, BA; Janet Miller, PhD; Angie Fedele, BA; Jack Collins, MBA; Karen Smith, BS; Linda Lotspeich, MD; Lisa A. Croen, PhD; Sally Ozonoff, PhD; Clara Lajonchere, PhD; Judith K. Grether, PhD; Neil Risch, PhD*

Concordance in male twin pairs:	Dizygotic	Monozygotic
ASD (n = 90 pairs)	.31	.77
Autism (n = 71 pairs)	.21	.58

## Antidepressant Use During Pregnancy and Childhood Autism Spectrum Disorders

*Lisa A. Croen, PhD; Judith K. Grether, PhD; Cathleen K. Yoshida, MS; Roxana Odouli, MSPH; Victoria Hendrick, MD*

Birth cohort 1995 – 1999, (n = 88,163)  
 Any antidep 1 yr prior to delivery:  
 20/298 (6.7%) ASD vs 50/1507 (3.3%) con.  
 1<sup>st</sup> trimester SSRI use : 14/298 ASD vs 19/1507 con. (O.R. = 3.8)

# What caused this to happen and can it be prevented?

## PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

Volume 128, Number 2, August 2011

### Perinatal and Neonatal Risk Factors for Autism: A Comprehensive Meta-Analysis

**AUTHORS:** Hannah Gardener, ScD,<sup>a</sup> Donna Spiegelman, ScD,<sup>a,b</sup> and Stephen L. Buka, ScD<sup>c</sup>

40 studies, > 60 variables:  
Low birth wt  
Resp distress  
Med prob < 1 mo

*Epidemiology* • Volume 22, Number 4, July 2011

### Prenatal Vitamins, One-carbon Metabolism Gene Variants, and Risk for Autism

*Rebecca J. Schmidt,<sup>a,b</sup> Robin L. Hansen,<sup>b,c</sup> Jaana Hartiala,<sup>d</sup> Hooman Allayee,<sup>d</sup> Linda C. Schmidt,<sup>e</sup> Daniel J. Tancredi,<sup>c,f</sup> Flora Tassone,<sup>b,e</sup> and Irva Hertz-Picciotto<sup>a,b</sup>*

### Month of Conception and Risk of Autism

*Ousseny Zerbo,<sup>a</sup> Ana-Maria Iosif,<sup>a</sup> Lora Delwiche,<sup>a</sup> Cheryl Walker,<sup>a</sup> and Irva Hertz-Picciotto,<sup>a,b</sup>*

# What caused this to happen and can it be prevented?

Neuron

## Article

Neuron 70, 863–885, June 9, 2011

### Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism

Stephan J. Sanders,<sup>1,2,3,4</sup> A. Gulhan Ercan-Sencicek,<sup>1,2,3,4</sup> Vanessa Hus,<sup>5,36</sup> Rui Luo,<sup>6,36</sup> Michael T. Murtha,<sup>1</sup> Daniel Moreno-De-Luca,<sup>7</sup> Su H. Chu,<sup>8</sup> Michael P. Moreau,<sup>9</sup> Abha R. Gupta,<sup>2,10</sup> Susanne A. Thomson,<sup>11</sup> Christopher E. Mason,<sup>12</sup> Kaya Bilguvar,<sup>1,4,13</sup> Patricia B.S. Celestino-Soper,<sup>14</sup> Murim Choi,<sup>4,27</sup> Emily L. Crawford,<sup>15</sup> Lea Davis,<sup>15</sup> Nicole R. Davis Wright,<sup>2</sup> Rahul M. Dhodapkar,<sup>2</sup> Michael DiCola,<sup>9</sup> Nicholas M. DiLullo,<sup>2</sup> Thomas V. F. Fielding-Singh,<sup>16</sup> Daniel O. Fishman,<sup>17</sup> Stephanie Frahm,<sup>9</sup> Rouben Garagaloyan,<sup>18</sup> Gerald S. Goh,<sup>4</sup> Sindhuja Kammela,<sup>2</sup> Lambertus Klei,<sup>19</sup> Jennifer K. Lowe,<sup>20</sup> Sabata C. Lund,<sup>5</sup> Anna D. McGrew,<sup>11</sup> Kyle A. Meyer,<sup>2</sup> William J. Moffat,<sup>2</sup> John D. Murdoch,<sup>4</sup> Brian J. O’Roak,<sup>22</sup> Gordon T. Ober,<sup>2</sup> Rebecca S. Pottenger,<sup>23</sup> Melanie J. Youeun Song,<sup>2</sup> Qi Wang,<sup>9</sup> Brian L. Yaspan,<sup>11</sup> Timothy W. Yu,<sup>24</sup> Ilana R. Yurkiewicz,<sup>2</sup> Arthur L. Beaudet,<sup>14</sup> Rita M. Cantor,<sup>6,25</sup> Martin Curland,<sup>18</sup> Dorothy E. Grice,<sup>26</sup> Murat Günel,<sup>1,4,13</sup> Richard P. Lifton,<sup>4,27</sup> Shrikant M. Donna M. Martin,<sup>29</sup> Chad A. Shaw,<sup>14</sup> Michael Sheldon,<sup>30</sup> Jay A. Tischfield,<sup>30</sup> Christopher A. Walsh,<sup>31</sup> Eric M. David H. Ledbetter,<sup>33</sup> Eric Fombonne,<sup>34</sup> Catherine Lord,<sup>5,35</sup> Christa Lese Martin,<sup>7</sup> Andrew I. Brooks,<sup>9</sup> James S. Edwin H. Cook, Jr.,<sup>15,36</sup> Daniel Geschwind,<sup>20,36</sup> Kathryn Roeder,<sup>8</sup> Bernie Devlin,<sup>19</sup> and Matthew W. State<sup>1,2,3</sup>

Simons Simplex Study:  
8% with CNVs  
1 in 38 with recurrent  
5 recurrent mutations  
Synaptic genes implicated

### Rare De Novo and Transmitted Copy-Number Variation in Autistic Spectrum Disorders

Neuron 70, 886–897, June 9, 2011

Dan Levy,<sup>1</sup> Michael Ronemus,<sup>1</sup> Boris Yamrom,<sup>1</sup> Yoon-ha Lee,<sup>1</sup> Anthony Leotta,<sup>1</sup> Jude Kendall,<sup>1</sup> Steven Marks,<sup>1</sup> B. Lakshmi,<sup>1,4</sup> Deepa Pai,<sup>1,5</sup> Kenny Ye,<sup>2</sup> Andreas Buja,<sup>3</sup> Abba Krieger,<sup>3</sup> Seungtae Yoon,<sup>1,6</sup> Jennifer Troge,<sup>1</sup> Linda Rodgers,<sup>1</sup> Ivan Iossifov,<sup>1</sup> and Michael Wigler<sup>1,\*</sup>

### Rare De Novo Variants Associated with Autism Implicate a Large Functional Network of Genes Involved in Formation and Function of Synapses

Sarah R. Gilman,<sup>1,3</sup> Ivan Iossifov,<sup>2,3,\*</sup> Dan Levy,<sup>2</sup> Michael Ronemus,<sup>2</sup> Michael Wigler,<sup>2</sup> and Dennis Vitkup<sup>1,\*</sup>

Neuron 70, 898–907, June 9, 2011



# Where can I turn for services?

Matern Child Health J

Published online: 11 June 2011

## Unmet Need and Problems Accessing Core Health Care Services for Children with Autism Spectrum Disorder

Giuseppina Chiri · Marji Erickson Warfield



Disability and Health Journal 4 (2011) 143–152

Disability and  
Health Journal

[www.disabilityandhealthjnl.com](http://www.disabilityandhealthjnl.com)

Research Papers

### Parenting aggravation and autism spectrum disorders: 2007 National Survey of Children's Health

Laura A. Schieve, Ph.D.<sup>a,\*</sup>, Sheree L. Boulet, Dr.PH.<sup>a</sup>, Michael D. Kogan, Ph.D.<sup>b</sup>,  
Marshalyn Yeargin-Allsopp, M.D.<sup>a</sup>, Coleen A. Boyle, Ph.D.<sup>a</sup>, Susanna N. Visser, M.S.<sup>a</sup>,  
Stephen J. Blumberg, Ph.D.<sup>c</sup>, Catherine Rice, Ph.D.<sup>a</sup>

<sup>a</sup>National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA 30333, USA

<sup>b</sup>Maternal and Child Health Bureau, Health Research and Services Administration, Rockville, MD 20857, USA

<sup>c</sup>National Center for Health Statistics, Centers for Disease Control and Prevention, Hyattsville, MD 20782, USA

# What does the future hold, particularly for adults?

J Autism Dev Disord  
DOI 10.1007/s10803-011-1291-0

ORIGINAL PAPER

## **Elderly with Autism: Executive Functions and Memory**

Hilde M. Geurts · Marlies E. Vissers

## **Do sheltered workshops enhance employment outcomes for adults with autism spectrum disorder?**


Robert Evert Cimera, Paul Wehman, Michael West and Sloane Burgess  
*Autism* published online 24 May 2011

## **College students on the autism spectrum**

*Prevalence and associated problems*

Susan W. White, Thomas H. Ollendick and Bethany C. Bray  
*Autism* published online 24 May 2011

# What other infrastructure and surveillance needs must be met?



Home | About NDAR | Data Sharing | Standards | Tools | Policy & Procedures | Training | FAQs

The National Database for Autism Research (NDAR) is a secure research data repository, which is funded by the National Institute of Health to accelerate scientific data sharing and collaboration among autism spectrum disorder (ASD) investigators.

- [Request Access](#) ⓘ
- [Contribute](#) ⓘ
- [Preview Data](#) ⓘ

Listed below are the projects that have contributed and are now sharing their data.

Research Spotlight
Preview Data

## Available Records: 106,210

Select Data Source(s): [Summary Data Approach - pdf](#)

NDAR       Pediatric MRI Data Repository

Select Gender: ▼      Select Phenotype: ▼      Select Sub Phenotype: ▼

Age From (in months):        Age To (in months):        Select Verbal IQ: ▼      Select Non-Verbal IQ: ▼

Available Data Type(s)

Clinical Assessments	Imaging	Genomics
Aberrant Behavior Checklist - Community <span style="float: right;">544</span>	MR Spectroscopy <span style="float: right;">1582</span>	Drug metabolism analysis / microarray
Autism Diagnostic Interview - Revised (ADI-R) - (2003) <span style="float: right;">1578</span>	MRI <span style="float: right;">1561</span>	Gene Regulation / microarray <span style="float: right;">820</span>
Autism Diagnostic Interview-Rev (ADI-R) Toddler 2004 <span style="float: right;">47</span>	MRI Anatomical <span style="float: right;">40</span>	Gene expression / PCR array
Autism Diagnostic Interview-Rev (ADI-R) Toddler 2006 <span style="float: right;">47</span>	MRI DTI <span style="float: right;">124</span>	Gene expression / microarray
Autism Diagnostic Observation Schedule (ADOS) Toddler <span style="float: right;">704</span>	fMRI <span style="float: right;">29</span>	Gene expression / sequencing
Autism Diagnostic Observation Schedule (ADOS)-Module 2 <span style="float: right;">403</span>		Gene expression microarray <span style="float: right;">3</span>
Autism Diagnostic Observation Schedule (ADOS)-Module 3 <span style="float: right;">780</span>		Gene regulation / PCR array
Autism Diagnostic Observation Schedule (ADOS)-Module 4 <span style="float: right;">264</span>		Gene regulation / mass spectrometry
Autism Diagnostic Observation Schedule(ADOS)-Module 1 <span style="float: right;">811</span>		Gene regulation / microarray
Autism Quotient Adult <span style="float: right;">12</span>		Gene regulation / sequencing

## Available Records: 106,210