Autism Research Update

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National Institutes of Health (NIH)

National Autism Conference

State College, Pennsylvania

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Presentation Outline

• NIH ACE Centers and Networks embarking on critical projects (minimal verbal ASD, females with ASD, diverse population)
• Changing demographics on individuals with ASD according to new CDC prevalence data
• IACC released 2013 strategic plan
• NIH workshop exploring mental health needs in IDDs
ACE Program

NIH awards $100 million for Autism Centers of Excellence Program
ACE Centers

- Susan Bookheimer, Ph.D. UCLA
  Biomarkers of Developmental Trajectories and Treatment in ASD
- Ami Klin, Ph.D. Emory University,
  Mechanisms of Risks and Resilience in ASD: Ontogeny, Phylogeny and Gene Disruption
- Helen Tager-Flusberg, Ph.D.
  Boston University
  Minimally Verbal ASD: From Basic Mechanisms to Innovative Intervention

ACE Networks
NIH ACE Centers and Networks embarking on critical projects

• Minimally verbal ASD:
  Who are these children? How can we assess their skills and knowledge? What treatment or interventions are effective?
  
• Females with ASD:
  Is ASD underdiagnosed in females? What accounts for the gender gap?

• ASD from diverse background:
  Can the current genetic findings in ASD be generalized to individuals of African descent?

Minimally Verbal School-Aged Children with ASD: the Neglected End of the Spectrum

• Two review articles (Autism Research 2013):
  stemming from NIH Workshop in 2010
  
• Highly heterogeneous group
  
• Need for novel technologies for assessing receptive language and cognition
  
• About 30% of individuals with ASD
  
• Fully nonverbal individuals are rare
ACE Centers
Helen Tager–Flusberg, Ph.D.
Boston University
Minimally Verbal ASD: From Basic Mechanisms to Innovative Interventions

- Examine heterogeneous phenotypes of a subset of ASD who fail to acquire spoken language
- Develop methods for assessing cognition, language, and behavior
- RCT- Auditory-Motor Mapping Training (AMMT)
- Study Speech mechanisms using a neurocomputational model of speech production and imaging
- Study auditory processing focusing on the segregation of inputs into meaningful units

Adaptive Interventions for Minimally Verbal Children with ASD in the Community
(Connie Kasari, Ph.D., Principal Investigator)
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- Defining response and measuring change in minimally verbal children (up to 20 words)
- Using a sequential intervention approach—individualized, considering early successes, adapting to indications of slow response
- Study the effects of adding parent training for early responders
- Two intervention methods—JASP-EMT (Joint Attention, Symbolic Play-Enhanced Milieu Teaching) & CORE-DDT (Discrete Trial Training)

Multimodal Developmental Neurogenetics of Females with ASD
(Kevin Pelphrey, Ph.D., Principal Investigator)
Multimodal Developmental Neurogenetics of Females with ASD
(Kevin Pelphrey, Ph.D., Principal Investigator)

- I in 42 boys and 1 in 189 girls were identified with ASD
- Aims to identify sex differences in behavior phenotypes, brain structure, connectivity, and temporal dynamics for children with ASD
- Uses samples in the Simons Simplex Collection—including 304 females and 2,114 males with autism
- Can sex-specific factors or patterns of gene expression be identified?

Autism Genetics, Phase II: Increasing Representation of Human Diversity
(Daniel Geschwind, M.D., Ph.D., Principal Investigator)
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- Build on earlier work finding genetic variants associate with autism susceptibility
- Plan to recruit African-American families with a child with ASD (N=600)
- Identify gene variants associated with autism in Americans with self-reported African ancestry
- Evaluate disparities in diagnosis and access to care

Changing Demographics on Individuals with ASD

CDC Released New Prevalence Data
Methodology:

- Eight-year-old children from 2010 Data
- In 11 of the 14 Autism and Developmental Disabilities Monitoring (ADDM) Network sites
- Record (health and special education) review by trained clinicians
- Identified children with an ASD diagnosis and documented behaviors consistent with ASD
- Based on DSM-IV-TR

ADDM Network Sites for 2010 Data
**Key Findings:**

- 1 in 68 children identified with ASD
- Great variability across sites
- 1 in 42 boys versus 1 in 189 girls
- 1 in 63 white versus 1 in 81 black versus 1 in 93 Hispanic children
- IQ<70 (31%), IQ=71-85 (23%), IQ>85 (46%)
- 72% with documented ASD diagnosis
- Average age of diagnosis: age 4 for AD & PDD, age 6 for Asperger Disorder

**Interagency Autism Coordinating Committee (IACC)**

Federal advisory committee that coordinates all efforts within the Department of Health and Human Services (HHS)
Interagency Autism Coordinating Committee (IACC) update

- Established in 2006--Combating Autism Act
- Reauthorized in 2011--Combating Autism Reauthorization Act (CARA), expiring on September 30, 2014
- Workshop on co-occurring conditions--September 23, 2014, Porter Neuroscience Center, NIH
- Workshop will focusing on psychiatric disorders, sleep and neurological disorders, metabolic and immune disorders

Autism Collaboration, Accountability, Research, Education and Support Act

- Or the Autism CARES Act of 2014
- A five-year reauthorization of autism research and screening programs
- Requiring a report to Congress on young adults with ASD and the transition from school-based services to services available to adults
- Requiring an official within DHHS to oversee national ASD research, services, and support activities
IACC Released the 2013 strategic plan update

- Encouraged more attention to the needs of adults with ASD
- More focus on the most disabled individuals and underserved populations
- Need for interventions for co-occurring conditions
- Developing efficacious, efficient, scalable, and cost-effective interventions, tools, and practices
- Need for interventions that can improve quality of life for individuals with ASD and their families

NIH Workshop:
Mental Health in Intellectual and Developmental Disabilities

Research Challenges and opportunities
June 2-3, 2014
Mental Health in IDDs Workshop - Background

• Funders: NICHD, NIMH, and Special Olympics International
• 2001 meeting “Emotional and Behavioral Health in Persons with MR/DD: Research Challenges and Opportunities”
• Higher prevalence than the general population (Anxiety, Depression, ADHD)
• Unrecognized, undiagnosed, and untreated
• Individuals with IDDs routinely excluded in NIH research

Mental Health in IDDs Workshop - What we know

• Rare IDD syndromes with known genetic causes are associated with mental health disorders (e.g., social anxiety and fragile X syndrome)
• ABA approach (single-case, functional analysis) effective for self-injury, aggression, and others
• Promising family-centered interventions
• Promising novel treatment strategies: multimodal treatments and technology-aided cognitive training
• Successful evidence-based services provision models
Mental Health in IDDs Workshop—what we need

• Better tools for diagnosis
• Outcome measures targeting phenotypic symptoms (e.g. irritability, repetitive behavior, mood instability) and fundamental process (e.g., memory, sleep/wake cycle)
• Methods to address heterogeneity issues
• Treatment that involves innovative technology
• Research addressing transition to adulthood, community integration, and family adversity
• Study designs and consent processes to allow inclusion of individuals with low IQs

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NIH Director’s Blog
Autism Architecture; Unrolling the Genetic Blueprint

• NIH-funded study published in Nature Genetics (J. Buxbaum et al July 2014)
• Large sample of individuals with autism (n=3000) from Sweden’s universal health registry
• Most genetic risk for autism resides with common variation

http://directorblog.nih.gov/

Thank you!

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