DSM-5
Diagnostic Approaches to Autism Spectrum Disorder

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Disclosures

• Research Support
  – Janssen – study completed
  – NIH

• Consulting
  – Janssen – non-pharmacological research project
What is a Neurodevelopmental Disorder? (1)

– Usually Syndromal
– Pediatric Onset
– Affecting Brain Function
– Clinical Effects on
  • Emotion
  • Cognition
  • Behavior
What is a Neurodevelopmental Disorder?

• Etiology
  – Usually a genetic component
  – Often Familial
  – Sometimes heritable
  – May be related to *de novo* events
  – Role for epigenetics
  – Role for Gene-Environment Interactions (GEX)
What is a Neurodevelopmental Disorder? (3)

per DSM5

1. Intellectual Disability (ID) (Intellectual Development Disorder)
2. Global Development Delay
3. Unspecified Intellectual Disability (Intellectual Developmental Disorder)
4. Language Disorder
5. Speech Sound Disorder
6. Childhood-Onset Fluency Disorder (Stuttering)
7. Social Pragmatic Communication Disorder (SCD)
8. Unspecified Communication Disorder
9. Autism Spectrum Disorder (ASD)
10. Attention Deficit Hyperactivity Disorder (ADHD)
11. Other Specified Attention-Deficit Hyperactivity Disorder
12. Unspecified Attention-Deficit Hyperactivity Disorder
13. Specific Learning Disorder
14. Developmental Coordination Disorder
15. Stereotypic Movement Disorder
16. Tic Disorder
17. Other Specified Tic Disorder
18. Unspecified Tic Disorder
19. Other Specified Neurodevelopmental Disorder
20. Unspecified Neurodevelopmental Disorder
What is a Neurodevelopmental Disorder?

Consider Other Neurodevelopmental Disorders:
- OCD
- Eating Disorders
- Mood Disorders
  - Bipolar Disorder
  - Major Depressive Disorder
- Substance Abuse
- Schizophrenia
- Trauma related disorders
- Epilepsy
- Alzheimer’s Disease
**Autism/ASD – A Syndrome**

- **Syndrome**
  - Group of symptoms that tend to cluster together and share a common natural history/course

- **Disease**
  - A syndrome for which there is either:
    - A known etiology (or cause)
    - A known pathophysiological process
    - Both

- **ASD is not so unusual because:**
  - Like the most medical conditions, ASD is a syndrome
Leo Kanner
(1894-1981)

“Autistic Disturbances of Affective Contact”

The Nervous Child, 1943
Autism Spectrum Disorders (ASD)

- The 2 “D’s” = Delay & Deviations

- Domains of Impairment
  a. Social/Communication Development
     • Joint Attention
     • Theory of Mind
     • Reciprocity
     • Expressive/Receptive Language
  b. Restricted/Repetitive & Stereotyped Patterns of Interests and Activities (including IS)
Autism & Autistic Spectrum Disorders (ASD)

• Course
  a. Consistent over time
  b. Some symptoms decrease
     a. Stereotypies
     b. Visual regard
  c. Some symptoms persist
     - Lack of social reciprocity
     - Language abnormalities
     - Restrictive and repetitive behaviors/IS
NSAC Criteria
1978

A. Disturbed rate &/or sequence of development
B. Disturbed response to sensory stimuli
C. Disturbed speech/language, cognitive development
D. Disturbed Quality to relate appropriately to people, events and objects
DSM-IV
Pervasive Developmental Disorders (PDD)

299.0  Autistic Disorder
299.8  Asperger’s Disorder
299.9  Rett’s Syndrome
299.1  Childhood Disintegrative Disorder
299.8  Pervasive Developmental Disorder, Not Otherwise Specified (PDD-NOS)
## DSM-IV Autism Spectrum Disorders

<table>
<thead>
<tr>
<th>Deficits</th>
<th>Reciprocal Social</th>
<th>Language</th>
<th>Cognitive</th>
<th>Repetitive, Restricted</th>
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</thead>
<tbody>
<tr>
<td>Autism</td>
<td>+</td>
<td>+</td>
<td>+/-</td>
<td>+</td>
</tr>
<tr>
<td>Aspergers</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td>PDD-NOS</td>
<td>+</td>
<td>+/-</td>
<td>+/-</td>
<td>+</td>
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</table>
DSM 5: ASD

A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays

B. Restricted, repetitive patterns of behavior, interests, or activities

C. Symptoms must be present in early childhood

D. Symptoms together limit and impair everyday functioning.
A. Delay and/or abnormal patterns of development before age 3
B. Qualitative impairment in reciprocal social interactions
C. Qualitative impairments in communication
D. Restricted, repetitive and stereotyped patterns of behavior, interests and activities

A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays
B. Restricted, repetitive patterns of behavior, interests, or activities
C. Symptoms must be present in early childhood
D. Symptoms together limit and impair everyday functioning.
Similar cross-over, for example:

A. Qualitative Impairments in Social Interaction >>>A

B. Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:

- encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus >>>B1
- apparently inflexible adherence to specific, nonfunctional routines or rituals >>>B2
- stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements) >>>>B1
- persistent preoccupation with parts of objects >>>B3
A. Social Communication Disorder (SCD) is
   - an impairment of **pragmatics**
   - diagnosed based on difficulty in the social uses of verbal and nonverbal communication

B. Low social communication abilities
   - result in functional limitations

C. Rule out Autism Spectrum Disorder

D. Symptoms must be present in early childhood
New Definition of Autism Will Exclude Many, Study Suggests

Mary Meyer, right, of Ramsey, N.J., said that a diagnosis of Asperger syndrome was crucial for her daughter, Susan, 37.

By BENEDICT CAREY
Published: January 19, 2012
### Prevalence Estimates: 
**DSMIV PDD, DSM5 ASD, DSM5 SCD**

<table>
<thead>
<tr>
<th></th>
<th>% DSMIV PDD</th>
<th>% DSM5 ASD</th>
<th>% DSM5 SCD</th>
<th>%DSM5 ASD+SCD</th>
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</thead>
<tbody>
<tr>
<td><strong>Population</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Total</td>
<td>2.64</td>
<td>2.20</td>
<td>0.49</td>
<td>2.69</td>
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<tr>
<td>GPS</td>
<td>1.89</td>
<td>1.46</td>
<td>0.49</td>
<td>1.95</td>
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<tr>
<td>Clinical</td>
<td>0.75</td>
<td>0.75</td>
<td>0.00</td>
<td>0.76</td>
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<tr>
<td><strong>DSMIV PDD</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Austistic Dis</td>
<td>1.04</td>
<td>1.03</td>
<td>0.001</td>
<td>1.04</td>
</tr>
<tr>
<td>Asperger</td>
<td>0.60</td>
<td>0.55</td>
<td>0.05</td>
<td>0.59</td>
</tr>
<tr>
<td>PDD-NOS</td>
<td>1.00</td>
<td>0.63</td>
<td>0.32</td>
<td>0.94</td>
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### DSMIV cases not meeting DSM5 Criteria

<table>
<thead>
<tr>
<th>Other Diagnoses</th>
<th>Divergent Group, n=22 (7.5%)</th>
<th>% of Divergent</th>
<th>% Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>SCD</td>
<td>14</td>
<td>64</td>
<td>4.8</td>
</tr>
<tr>
<td>SCD+other Psych Disorder</td>
<td>4</td>
<td>14</td>
<td>1.4</td>
</tr>
<tr>
<td>Other Psych Disorder</td>
<td>5</td>
<td>22</td>
<td>1.7</td>
</tr>
<tr>
<td>No Diagnosis</td>
<td>0</td>
<td>0</td>
<td>0</td>
</tr>
</tbody>
</table>
So, will people be left out?

- Will patients with DSM-IV Pervasive Developmental Disorder not meet DSM-5 criteria?
  - YES
    - A few ~ 3%
    - Rett’s Syndrome - All
    - Autistic Disorder - None
    - Aspergers - very few
      - > SCD
    - PDD-NOS
      - > SCD (most)
      - > Other Psychiatric Disorder - ? did not have PDD in the first place
        » ADHD & Anxiety Disorder
Sources of Diagnostic Information

- Screening
- Clinical Interview
  - History
  - Direct Observation
- Standard Diagnostics
  - ADI
  - ADOS
  - CARS
- Adaptive Function
  - Vineland
  - ABC
- Physical Examination
  - Neurological
  - Sensory Exam
- Laboratory
  - Only as indicated
Most children with ASD get better over time!

Our goals are:

Search for etiologies that can be used to

  Enhance treatment

  Prevent ASD

In the meantime:

  Use treatments that improve the rate at which individuals with ASD acquire skills and the ability to use those skills for independent and semi-independent living
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