DSM-5 and Autism
Changes are in the air

W. David Lohr, M.D.
Assistant Professor Child Psychiatry
Co-Clinical Director University of Louisville Autism Center
University of Louisville School of Medicine

wdlohr01@louisville.edu
502-852-6941
New Definition of Autism Will Exclude Many, Study Suggests

- “proposed changes in the definition of autism would sharply reduce the skyrocketing rate at which the disorder is diagnosed”
- “proposed changes would probably exclude people with a diagnosis who were higher functioning”
- “experts…strongly disagree”
  - “I don’t know how they’re getting those numbers”, Catherine Lord

- New York Times, Jan 19, 2012 by Benedict Carey
“New Definition of Autism Will Exclude Many, Study Suggests”

♦ “45% of children and adults with higher functioning autism will qualify for proposed DSM-5 criteria”

♦ Access to services may be threatened

♦ Increased clarity

• New York Times, Jan 19, 2012 by Benedict Carey
Autism Prevalence On The Rise*
There has been a 600% increase in prevalence over the last two decades.

AUTISM SPEAKS™
It’s time to listen.
www.AutismSpeaks.org

*Recent research has indicated that changes in diagnostic practices may account for at least 25% of the increase in prevalence over time, however much of the increase is still unaccounted for and may be influenced by environmental factors.
Prevalence continues to grow

- Current CDC estimates 1 in 88 children aged 8 years have an autism spectrum disorder
  - 1 in 54 boys
  - 1 in 252 girls


- Reasons for increase
  - Awareness
  - Newer community studies
  - Better methods
Definitions

- **Sensitivity**
  - Measures a test’s ability to find those patients with a disease
  - If a person has a condition, how often will the test find it
  - Finds true positives

- **Specificity**
  - Measures a test’s ability to identify those patients without a disease
  - If a person is healthy, how often will the test find it
  - Finds true negatives

  - [wikipedia.org](http://wikipedia.org)
<table>
<thead>
<tr>
<th>Expert Truth</th>
<th>AP Detection Algorithm</th>
<th>T</th>
<th>F</th>
</tr>
</thead>
<tbody>
<tr>
<td>T</td>
<td>TP</td>
<td></td>
<td>FN</td>
</tr>
<tr>
<td>F</td>
<td>FP</td>
<td></td>
<td>TN</td>
</tr>
</tbody>
</table>

Figure 2.4. Contingency table used in APCAT to score the fuzzy recognizer performance against the truth field defined by the human experts. The “TP” represents True Positive results, “FN” is False Negative results, “FP” is False Positive results, and “TN” is True Negative results.
Reliability vs. Validity

- Reliable Not Valid
- Low Validity Low Reliability
- Not Reliable Not Valid
- Both Reliable and Valid

by Experiment-Resources.com
History of DSM and autism

- Autism was first recognized in DSM-III
  - Monothetic (individuals had to meet all diagnostic criteria)
  - Classical autism
  - Emphasis on categories not etiology
History of DSM and autism

- DSM-III-R
  - Developmental orientation
  - Polythetic (individuals could meet a specific number of sub-criteria from a set of criteria)
  - Broader diagnostic group
Table 1. *Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR)*

**Diagnostic Criteria for Autistic Disorder**

A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):

1. Qualitative impairment in social interaction, as manifested by at least two of the following:
   a. Marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
   b. Failure to develop peer relationships appropriate to developmental level
   c. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by lack of showing, bringing, or pointing out objects of interest)
   d. Lack of social or emotional reciprocity

2. Qualitative impairments in communication as manifested by at least one of the following:
   a. Delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
   b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
   c. Stereotyped and repetitive use of language or idiosyncratic language
   d. Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level

3. Restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
   a. Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
   b. Apparent inflexibility adherence to specific, nonfunctional routines or rituals
   c. Stereotyped and repetitive motor manner (e.g., hand or finger flapping or twisting, or complex whole-body movements)
   d. Persistent preoccupation with parts of objects

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years:

1. Social interaction
2. Language as used in social communication
3. Symbolic imaginative play

C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder

From autism to Asperger’s Disorder
DSM-IV criteria for Asperger’s Disorder

- Impaired social interaction, with at least two of four criteria
- Restricted, repetitive, and stereotypical behaviors and interests with at least one of four criteria
- Significant impairment in important areas of function
- No overall delay in language
- No significant delay in cognitive development, (mental retardation), or in adaptive skills
- Criteria are not met for autism or schizophrenia
Proposed DSM-5 criteria for Autism Spectrum Disorder, revised Jan 26, 2011

• Must meet criteria A, B, C, and D:

• A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, and manifest by all 3 of the following:
  • 1. Deficits in social-emotional reciprocity
  • 2. Deficits in nonverbal communicative behaviors used for social interaction
  • 3. Deficits in developing and maintaining relationships, appropriate to developmental level (beyond those with caregivers)

• www.dsm5.org/ProposedRevisions/Pages/proposedrevision.aspx?rid=94#
Proposed DSM-5 criteria for Autism Spectrum Disorder, revised Jan 26, 2011

B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least two of the following:

1. Stereotyped or repetitive speech, motor movements, or use of objects
2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change
3. Highly restricted, fixated interests that are abnormal in intensity or focus
4. Hyper-or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment

www.dsm5.org/ProposedRevisions/Pages/proposedrevision.aspx?rid=94#
C. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)

D. Symptoms together limit and impair everyday functioning.

www.dsm5.org/ProposedRevisions/Pages/proposedrevision.aspx?id=94#
<table>
<thead>
<tr>
<th>Severity level for ASD</th>
<th>Social communication</th>
<th>Restricted interests and repetitive behavior</th>
</tr>
</thead>
<tbody>
<tr>
<td>Level 3 (requiring very substantial support)</td>
<td>Severe deficits with very limited initiation of social interactions and minimal response to social overtures from others.</td>
<td>markedly interfere with functioning in all spheres. Marked distress, very difficult to redirect.</td>
</tr>
<tr>
<td>Level 2 (requiring substantial support)</td>
<td>Marked deficits with limited initiation of social interactions and reduced or abnormal response to social overtures from others.</td>
<td>obvious interference with functioning, Distress or frustration is apparent, difficult to redirect</td>
</tr>
<tr>
<td>Level 1 (requiring support)</td>
<td>Without supports in place, deficits in social communication cause noticeable impairments.</td>
<td>significant interference with functioning in one or more contexts. Resists interruption or redirection</td>
</tr>
</tbody>
</table>
Summary of DSM-5 changes for Autism

♦ Autism, Asperger’s disorder, and PDD-NOS will be combined into a single category

♦ Monothetic for social-communication symptoms
  ♦ Combines DSM-IV criteria for social interaction and impairments of communication

♦ Polythetic for restricted, repetitive behaviors
  ♦ 2 of 4 symptoms must be present
  ♦ Sensory symptoms are included

♦ Universal age onset by early childhood
DSM-5 diagnostic simplicity

DSM-IV
2,027 different combinations

DSM-5 only has 11 different ways to meet diagnosis

McPartland JAACAP, 2012;51(4)368-383.
Problems with DSM-IV and autism

♦ Reliability
  ♦ Best-estimate clinical diagnoses compared to those using ADI-R and ADOS
  ♦ Experts differ on how they interpreted DSM-IV-TR data
    ♦ Regional differences
    ♦ Influenced by verbal IQ and language
  ♦ DSM-IV is difficult to implement
Problems with DSM-IV and autism

- **Validity**  
- Asperger’s disorder may not be distinct from high-functioning autism  
  Measurement of language delays by age 3 years
  - No effect on outcome
  - Difficult to retrospectively measure
- Limited neuropsychological distinctions from autism
- Precedence rule
- **PDD-NOS**
  - Residual category
  - Poorly defined, poor reliability of diagnosis
Goals for DSM-5 and autism

- Recognize “essential shared features” of ASD
  - Clearer, simpler diagnosis
  - Failure of social communication development

- Combine categorical and dimensional measures
  - Individualized diagnosis

- Reintegrate Asperger’s disorder and PDD-NOS into ASD
DSM-5 Neurodevelopmental Disorders Workgroup

- 13 members chaired by Susan Swedo, M.D.
  - Chief, Pediatrics & Developmental Neuroscience Branch, NIMH

- Over 6,000 hours of meetings and teleconferences
  - Formulate draft criteria
  - Field trials and data collection
  - Revision of draft criteria due Spring 2012
  - Open public feedback
  - Final draft data due November 30, 2012 with publication date of December 31, 2012
DSM-IV-TR vs. DSM-5

• 5,484 8-year olds in Finland rated for autism
  • Prevalence rate of ASD was 1 in 119, autism 1 in 244 via DSM-IV
  • DSM-5 less sensitive for Asperger’s disorder and high-functioning autism
    • 46% identification rate in those with an IQ > 50.
    • None of the 11 patients with Asperger’s were seen
DSM-IV-TR vs. DSM-5

- DSM-5 had Improved “construct validity” over DSM-IV-TR
  - Stereotyped and repetitive use of language
  - Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
  - Hyper-or hypo-reactivity to sensory input or unusual interest in sensory aspect of environment
DSM-5 validated


- 14,744 siblings from the Interactive Autism Network
- Hybrid model of categorical and dimensional criteria supported

<table>
<thead>
<tr>
<th></th>
<th>DSM-IV-TR</th>
<th>DSM-5</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sensitivity</td>
<td>0.95</td>
<td>0.81</td>
</tr>
<tr>
<td>Specificity</td>
<td>0.86</td>
<td>0.97</td>
</tr>
</tbody>
</table>

- “superior specificity”
- At risk for false negative
  - Females
  - Asperger’s disorder
Sensitivity and Specificity of DSM-5 criteria

McPartland et al. JAACAP, 2012;51(4)368-383.

- 933 patients evaluated from DSM-IV field trial exposed to DSM-5 criteria
- Overall 60% of cases with ASD met revised DSM-5 criteria
- Overall specificity was 94.9%

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Sensitivity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autism</td>
<td>0.76</td>
</tr>
<tr>
<td>Asperger’s disorder</td>
<td>0.25</td>
</tr>
<tr>
<td>PDD-NOS</td>
<td>0.28</td>
</tr>
<tr>
<td>IQ &lt; 70</td>
<td>0.70</td>
</tr>
<tr>
<td>IQ &gt; 70</td>
<td>0.46</td>
</tr>
</tbody>
</table>
Social Communication Disorder

- Impairment of pragmatics diagnosed on difficulties in social use of verbal and non-verbal behavior
  - Not explained by learning disorders or IQ
- Excludes Autism Spectrum Disorder (RRB criteria)
- Onset in early childhood
SCD = ASD-RRB

- Social Communication Disorder = Autism Spectrum Disorder – restricted, repetitive behavior
- PDD-NOS?
- Operational criteria
**Proposed Modifications to DSM-5**

<table>
<thead>
<tr>
<th>DSM-5 draft criteria</th>
<th>Suggested changes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Must meet all 3 criteria for social communication and interaction</td>
<td>May meet 2 of 3 criteria for social communication and interaction</td>
</tr>
<tr>
<td>“deficits in nonverbal communication”</td>
<td>“deficits in verbal and/or nonverbal communication”</td>
</tr>
<tr>
<td>“Excessive adherence to routines and rituals”</td>
<td>“excessive adherence to routines and/or rituals”</td>
</tr>
<tr>
<td>“symptoms must be present in early childhood”</td>
<td>“symptoms present in childhood”</td>
</tr>
</tbody>
</table>

Increased sensitivity with modification

<table>
<thead>
<tr>
<th>Disorder</th>
<th>DSM-5 criteria</th>
<th>Modified criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autism Spectrum Disorders, FSIQ &gt; 50</td>
<td>46%, (n=26)</td>
<td>96%</td>
</tr>
<tr>
<td>High functioning autism (FSIQ &gt; 70)</td>
<td>73%, (N=11)</td>
<td>100%</td>
</tr>
<tr>
<td>Asperger’s disorder</td>
<td>0%, (n=11)</td>
<td>91%</td>
</tr>
</tbody>
</table>

Proposed Modifications to DSM-5

- Relaxing DSM-5 criteria with one less SCI or RRB criterion may increase sensitivity by 11% to 12%
  - Useful impact on those with Asperger’s disorder
  - Patients with limited early childhood history

- “symptoms must be present in early childhood” should be modified to allow children or adults without accurate early history can be diagnosed
Proposed Modifications to DSM-5

- Relax SCI criteria to allow 2 of 3 criteria to be used
- AND Lower RRB threshold from 2 to 1
  - McPartland JAACAP, 2012;51(4)368-383.

<table>
<thead>
<tr>
<th></th>
<th>DSM-5</th>
<th>Relaxed SCI criteria</th>
<th>AND Lowered RRB threshold</th>
</tr>
</thead>
<tbody>
<tr>
<td>sensitivity</td>
<td>0.61</td>
<td>0.75</td>
<td>0.91</td>
</tr>
<tr>
<td>specificity</td>
<td>0.95</td>
<td>0.85</td>
<td>0.75</td>
</tr>
</tbody>
</table>
Finding the right balance
Pros of DSM-5 and autism

- Improved specificity
  - Stability
  - Clarity

- Dimensional approach
  - Core issue of autism = social communication deficits
    - Genetic influence
    - Normal distribution in population

- Measures of severity
Pros of DSM-5 and autism

- Increased homogeneity
- Longitudinal research
- Endophenotypes
- Genotypes
Cons of DSM-5 and autism

- Decreased sensitivity
  - Those with high functioning autism and Asperger’s disorder may not meet criteria for DSM-5
  - Social communication disorder

- Effect on service eligibility

- Compatibility with prior research subtypes

- Fit with ICD-11
Future Directions

- Draft revisions
- Prospective field studies
  - Phase II testing
  - Compare DSM-5 vs. DSM-IV and DSM-5 vs. expert clinical diagnosis
- Define social communication disorder
Future Directions

- Search domains of social communication and repetitive, restricted behaviors
  - Isolate endophenotypes
  - Find biologic and genetic markers

- Suitable instruments to measure social communicative function
The nature of autism

♦ Lumpers
  ♦ Population distribution of social function
  ♦ Rates in twins and siblings

♦ Splitters
  ♦ Latent symptoms
  ♦ Brain development

♦ Hybrid model
  ♦ Supported by recent validity studies by Mandy et al. and Frazier et al.
FRAP!

Frenetic
random
acts of
play

W. David Lohr, M.D.

Assistant Professor Child Psychiatry

Co-Clinical Director University of Louisville Autism Center

University of Louisville School of Medicine

wdlohr01@louisville.edu

502-852-6941