DSM-5:
The New Diagnostic Criteria For Autism Spectrum Disorders

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Consultation/Advisory Boards
(More than 1 year ago)

• Eisai
• Novartis U.S.A.
• Novartis Global
DSM-5: The New Criteria

What?
DSM-5 will be released in May 2013
Book, CD, website

Single diagnosis:
Autism Spectrum Disorder (ASD)
Concerns about ASD in DSM-5

- Sensitivity has been “sacrificed” in order to improve specificity
  - Social communication domain
  - Restrictive interests and repetitive behaviors domain
- Merging Asperger disorder (and PDD-NOS) into autism spectrum disorder results in loss of identity and ignores uniqueness of Asperger dx
- Pre-/post DSM-5 research studies will not be comparable
- Changes in criteria threaten services delivery
History of DSM-5 & Autism

DSM-I (1952) & DSM-II (1968)
No term Autism or Pervasive Developmental Disorder
Closest term: Schizophrenic Reaction (Childhood Type)

1980 DSM-III
Pervasive Developmental Disorders (PDD):
Childhood Onset PDD, Infantile Autism, Atypical Autism

1987 DSM-III-R
Pervasive Developmental Disorders (PDD):
PDD-NOS, Autistic Disorder

1994 DSM-IV
Pervasive Developmental Disorders (PDD):
PDD-NOS, Autistic Disorder, Asperger Disorder,
Childhood Disintegrative Disorder, Rett syndrome

2000 DSM-IV-TR
Same diagnoses, text correction for PDD-NOS
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Who?
DSM-5 Neurodevelopmental Disorders Workgroup (2007 – Present)

MEMBERS
- Gillian Baird*
- Ed Cook*
- Francesca Happe*
- James Harris*
- Walter Kaufmann*
- Bryan King*
- Catherine Lord*
- Joseph Piven*
- Rosemary Tannock
- Sally Rogers*
- Sarah Spence*
- Susan Swedo*
- (Fred Volkmar – resigned in ‘09)
- Amy Wetherby*
- Harry Wright

ADVISORS
- Jim Bodfish
- Martha Denckla
- Maureen Lefton-Grief
- Nickola Nelson
- Sally Ozonoff
- Diane Paul
- Eva Petkova
- Daniel Pine
- Alya Reeve
- Mabel Rice
- Joseph Sergeant
- Bennett & Sally Shaywitz
- Audrey Thurm
- Keith Widaman
- Warren Zigman
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HOW?
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Work Process of the DSM-5 Neurodevelopmental Disorders Workgroup

• Biweekly teleconferences, semi-annual in-person meetings and additional web conferences, other “meetings” as needed. >2,500 hours of work over the course of 5 years.
• Expert consensus supported by literature reviews, secondary data analyses and clinical evaluations.
• Vetted through public comments, presentations at scientific and advocacy meetings, review by leading experts (e.g. Sir Michael Rutter, members of INSAR) and advocacy group members (e.g., Asperger groups)
• ALL autism groups now support the criteria set. Only GRASP and New England Asperger Support group continue to object (to removal of Asperger label)
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Why?
Concerns about application of DSM-IV

• Validity of the Pervasive Developmental Disorders category

• Consistency of some diagnoses (e.g., high-functioning autistic disorder vs. Asperger)

• Appropriateness of the use of certain diagnoses (e.g., PDD-NOS as mild neurodevelopmental disorder, Asperger as “odd” behaviors)

• Validity of some diagnoses (e.g., childhood disintegrative disorder)
“Autism” in DSM-IV

• Pervasive Developmental Disorders
  – Autism
  – Asperger disorder (NEW in DSM-IV)
  – Rett syndrome (New in DSM-IV)
  – Childhood disintegrative disorder (CDD) (New in DSM-IV)
  – PDD-NOS (Pervasive Developmental Disorder – Not Otherwise Specified) (New description in DSM-IV)
“Autism” in the Field

• DSM-IV is NOT the “Gold Standard”
• Autism Diagnostic Interview-Revised (Rutter & Lord) is used by researchers and academic centers
• Clinicians use ICD “autism” criteria of social deficits plus RRB’s
• CDC reports “autism” rates using all PDD categories
PDD-NOS in DSM-IV
(Actually, PDD-NVS for “Not Very Specific”)

“This category should be used when there is severe and pervasive development of reciprocal social interaction associated with impairment in either verbal or nonverbal communication skills OR with the presence of stereotyped behavior, interests and activities, but the criteria are not met for a specific PDD… For example, this category includes “atypical autism” – presentations that do not meet the criteria for Autistic disorder because of late age at onset, atypical symptomatology, or SUBTHRESHOLD SYMPTOMATOLOGY.”
Current Concept of Autism

1. Behavioral disorder/syndrome

2. Multiple etiologies: genetic and environmental

3. Lifelong disorder
   a) Different appearance (e.g., peer interactions change throughout life)
   b) Importance of early diagnosis
   c) Need for sustained support

4. **Selective** or **greater** impairment in social interaction

5. Common use of Autism Spectrum Disorder, including Autistic Disorder and PDD-NOS
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DSM-5: Conceptual Framework

- Social Impairment
  - Repetitive Behaviors & Restricted Interests
- Social Anxiety
- Epilepsy-EEG abnormalities
- Autism Spectrum Disorder
  - Speech/Communication Deficits
  - Repetitive Behaviors & Restricted Interests
- Intellectual Disability
  - Language Disorders
- ADHD
- Aggression

- Sleep Disturbance
- Motor problems
- Gastro-intestinal Dysfunction
- Immune Dysfunction
- OCD
Proposed Changes: Name of Category

• Delete the term “Pervasive Developmental Disorders”
  – Symptoms are not pervasive – they are specific to social-communication domain plus restricted, repetitive behaviors/fixated interests
  – Overuse of PDD-NOS leads to diagnostic confusion (and may have contributed to autism “epidemic”)
  – Overlap of PDD-NOS and Asperger disorder
• Recommend new diagnostic category: “Autism Spectrum Disorder”
Proposed Changes: Deletion of Rett Syndrome as a specific ASD

- Rett will be removed as a separate disorder
  - **JUSTIFICATION:**
    - ASD behaviors are not particularly salient in Rett Syndrome patients except for brief period during development.
    - ASD are defined by specific sets of behaviors, not etiologies (at present) so inclusion of Rett Disorder is atypical.
    - Patients with Rett Syndrome who have autistic symptoms can still be described as having ASD, and clinicians should use the specifier “with known genetic or medical condition” to indicate symptoms are related to Rett.
Proposed Changes:
Deletion of Childhood Disintegrative Disorder

• New knowledge that developmental regression in ASD is a continuous variable, with wide range in the timing and nature of the loss of skills, as well as the developmental milestones that are reached prior to regression.

• Rarity of CDD diagnosis makes systematic evaluation difficult, but review of accumulated world’s literature shows that CDD has important differences from other ASD’s, including the acuity and severity of regression, as well as co-occurring physical symptoms, such as loss of bowel and bladder control. (Need to look for neurological disorder)
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Proposed Changes: Elimination of Asperger Disorder

• New diagnosis in DSM-IV with little difference from autism
• Criteria used in DSM-IV do not match the original cases described by Asperger (his cases meet autism criteria)
• No clinical or research evidence for separation of Asperger disorder from autism (High functioning autism = Asperger dx)
• Diagnostic biases apparent, with rich, white males receiving Asperger dx, while poorer, non-Caucasian populations receive PDD-NOS diagnosis (See site differences in CDC surveillance data)
Proposed Changes:
Merging of ASDs into a Single Diagnosis

- AUTISM, ASPERGER and PDD-NOS will be collapsed into a single dx: AUTISM SPECTRUM DISORDER
  - JUSTIFICATION:
    - Scientific evidence and clinical practice show that a single spectrum better reflects the symptom presentation, time-course and response to treatment
    - Separation of ASD from typical development is reliable & valid while separation of disorders within the spectrum is not (e.g., Asperger and PDD-NOS used interchangeably, as are HFA and Asperger)
    - Many states provide services only for dx of autism; as expected, PDD-NOS and Asperger disorder are rare dx’s in those jurisdictions
Data from Simons Simplex Collection

N = sample size
F = % Females
A = Mean Age
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Data from Simons Simplex Collection

Diagram showing social affect distribution across different sites.
Single Spectrum but Significant Individual Variability

- Severity of ASD Symptoms
- Pattern of Onset and Clinical Course
- Etiologic factors
- Cognitive abilities (IQ)
- Associated conditions

CLINICIANS WILL BE ENCOURAGED TO DESCRIBE THESE DETAILS WITH DIAGNOSTIC SPECIFIERS
Proposed Changes: Number of Symptom Domains

• THREE will become TWO
  – Social Communication domain will be created by merger of key symptoms from the DSM-IV Social and Communication domains
  – Fixated interests and repetitive behavior or activity

JUSTIFICATION:

• Deficits in communication are intimately related to social deficits. The two are “manifestations” of a single set of symptoms that are often present in differing contexts.
• This de-emphasizes language skills not employed in the context of social communication.
• Fixes the “double-counting” problem of DSM-IV
DSM-5 Criteria for Autism Spectrum Disorder

Currently, or by history, 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

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;

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.
From DSM-IV to DSM-5

**DSM-IV**

A. Impairment in social interaction as manifested by at least one of the following:

1. 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.
2. Failure to develop peer relationships appropriate to developmental level.
3. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest).
4. Lack of social or emotional reciprocity.

**DSM-5**

A. Persistent deficits in social communication and interaction across multiple contexts, as manifested by at least two of the following:

1. Deficits in social-emotional reciprocity, which may range for example, from abnormal social approach and failure of normal back and forth conversation, to reduced sharing of interests, emotions, or affect, to failure to initiate or respond
2. Deficits in communicative behaviors used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication, to abnormalities in eye contact and body language or deficits in understanding and use of gestures, to a total lack of facial expressions and nonverbal interaction.
3. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts, difficulties in sharing imaginative play or in making friends, to absence of interest in peers.

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

1. Stereotyped or repetitive motor movements, use of objects, or speech (such as simple motor stereotypes, lining up toys or flipping plates, echolalia, idiosyncratic phrases).
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (such as extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food every day).
3. Highly restricted, fixated interests that are abnormal in intensity or focus (such as strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).
4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of the environment (such as apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smeling or touching of objects, visual fascination with lights or movement).

Becomes a specifier.
Examples of DSM-IV vs. DSM-5

For subcriterion A.3,

DSM-IV checklist item is “failure to develop peer relationships and abnormal social play.”

DSM-5 recommendations include higher-order impairments of “difficulties adjusting behavior to suit different social contexts.”
For criterion C, DSM-IV requires that symptoms begin prior to the age of 3 years.

DSM-5 requires that symptoms begin in early childhood, with the caveat that “symptoms may not be fully manifest until social demands exceed capacity” (during middle-school years, later adolescence, or young adulthood).
Other Changes for ASD

• Inclusion of Specifiers, such as:
  – “Associated with Known Medical or Genetic Condition or Environmental Factor” (e.g., Fragile X, VCFS, intrauterine valproate exposure)
  – Verbal abilities
  – Cognitive abilities
  – Severity of symptoms in the two domains
• Text description to include symptoms unique to various ages/developmental stages and verbal abilities
Social Communication Disorder

A. Persistent difficulties in the social use of verbal and nonverbal communication as manifest by deficits in the following:

   1) Using communication for social purposes, such as greeting and sharing information, in a manner that is appropriate for the social context;

   2) Changing communication to match context or the needs of the listener, such as speaking differently in a classroom than on a playground, communicating differently to a child than to an adult, and avoiding use of overly formal language.

   3) Following rules for conversation and storytelling, such as taking turns in conversation, rephrasing when misunderstood, and knowing how to use verbal and nonverbal signals to regulate interaction;

   4) Understanding what is not explicitly stated (e.g. inferencing) and nonliteral or ambiguous meanings of language, for example, idioms, jokes, metaphors and multiple meanings that depend on the context for interpretation.

B. Deficits result in functional limitations in effective communication, social participation, social relationships, academic achievement, or occupational performance.

C. Onset in the early developmental period (but deficits may not become fully manifest until social communication demands exceed limited capacities).

D. Deficits are not better explained by low abilities in the domains of word structure and grammar, or by intellectual disability, global developmental delay, Autism Spectrum Disorder, or another mental or neurologic disorder.
Social Communication Disorder

- Diagnosis is needed for:
  - Children with current dx of PDD-NOS on the basis of social communication deficits
  - Individuals with significant social skills deficits
- Criteria appeared to function well in field trials
- Should NOT be included in ASD section because it defines a group of individuals with related, but separate symptoms
## Sensitivity and Specificity of DSM-5

<table>
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<tr>
<th>DSM-5 FIELD TRIALS</th>
<th>Completed V1</th>
<th>Completed V2</th>
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<td><strong>Stanford</strong></td>
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<td><strong>The Children's Hospital</strong></td>
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<td><strong>Baystate Medical Center</strong></td>
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<td>145</td>
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<td><strong>Pediatric Sites</strong></td>
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<td>606 (293)</td>
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High reliability, highest among pediatric DSM-5
## Sensitivity and Specificity of DSM-5

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<tr>
<th></th>
<th>NONE</th>
<th>Autistic Disorder</th>
<th>Asperger Disorder</th>
<th>PDD-NOS</th>
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<tr>
<td>No DSM-5 ASD/SCD</td>
<td>185</td>
<td>0</td>
<td>3 (14%)</td>
<td>5 (22%)</td>
<td></td>
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NOTE: Some DSM-IV cases “lost” their diagnosis with DSM-5 criteria, but overall, there was an increase in ASD cases. Further, when SCD cases are added, there was a 14% increase in new cases of ASD/SCD.
Sensitivity, Specificity, Reliability & Validity
DSM-5 ASD Criteria

Specificity and Sensitivity of DSM-5 ASD criteria in Fields Trials are comparable to DSM-IV

NDD Workgroup has begun reviewing archived patient interviews to assess Validity of DSM-5 Criteria and areas for improvement

Additional Prospective studies are required.
Retrospective data are NOT useful for determining utility of DSM-5

Non-DSM Measures of Social Interaction (SRS, ABC-C)?
Decision to include Asperger Syndrome & PDD-NOS within one ASD diagnosis

- Lack of specificity and sensitivity in separating Asperger from “high functioning autism”
- Lack of accurate historical information about early language development put emphasis on current speech (trainable)
- Overlap of AUT and ASP when VIQ controlled
- Differential use of dx’s by race and SES status
- Consideration of access to services (Not provided for PDD-NOS or Asperger dx)
Criticisms of DSM-5 ASD Criteria

Research done prior to DSM-5 won’t be comparable with research done afterwards

- ASD is already preferred term for research (See p.108 of survey)
- ADI-R and ADOS are diagnostic standards for research

Scientific Publications Citing ASD v PDD in the Title

<table>
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<tr>
<th>Year</th>
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<th>PDD</th>
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<td>2006</td>
<td>49</td>
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<td>2012</td>
<td>197</td>
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Criticisms of DSM-5 ASD Criteria

“Changes in criteria threaten services delivery”

• Actually, a single diagnosis of ASD will IMPROVE access to services (See CPHC document)

• PDD-NOS and Asperger disorder do not qualify for services in 14 states

• Apparent biases in diagnostic labelling with rich, white males receiving (less-stigmatizing) dx of Asperger disorder, while poor, non-white males and females receive PDD-NOS (or autism)