One of our own

- Restricted area of interest
- Star Trek
- Borg
  - “Negotiation is irrelevant. You will be assimilated.”
How reliable is the diagnosis:

- “We scrapped these labels 18 years ago at our place, after two years of saying we were only going to take the so-called autistic, schizophrenic, psychotic, or symbiotic child. We found that all these labels are just meaningless. We learn more about these kids from working with them on a day-to-day basis. Too many people feel that sticking a label on them means that they now know what this kid needs. I think this is a dangerous, misleading and destructive process.”

Dr. Carl Fenichel at the Leo Kanner Colloquium on Child Development, Deviations and Treatment at Chapel Hill, NC, 1973
Wastebasket:

- “A tendency has arisen to set up a pseudo-diagnostic wastebasket into which as assortment of heterogeneous conditions were thrown indiscriminately. . . . Such looseness threw all curiosity about diagnostic criteria to the winds as irrelevant impediments on the road to therapy, which was applied to all-comers as if their problems were identical. The therapeutic cart was put before the diagnostic horse and, more often than not, the horse was left out altogether.”

Kanner addressing the abuses of the term “autism” in a lecture in NY in 1965.
DSM History


- DSM-I 1952
- DSM-II 1968
- DSM-III 1980
- DSM-III-R 1987
- DSM-IV 1994
- DSM-IV-TR 2000
- DSM-5 2013
Paradigm Shift?

• Initially claimed that the new DSM would be a paradigm shift

• Now:
  • A conservative document that will have minimal impact on diagnostic rates
Non-ASD Changes

- Hoarding
  - Now its own mental disorder

- Premenstrual dysphoric disorder
  - Severe form of premenstrual syndrome

- Binge Eating Disorder
  - Severe form of gluttony
Thorny Changes (Non-ASD)

- Depression
  - Eliminate bereavement exclusion
  - Grieving now counts
- Bipolar
  - Wanted to slow down diagnosing kids
  - Settled on Disruptive Mood Dysregulation Disorder
  - Like ADHD with explosive features
  - Careful not to medicalize frequent temper tantrums
Changes in ASD in the DSM-5

- Single diagnostic category: PDD $\rightarrow$ ASD
  - No more Autistic Disorder, Asperger’s, or PDD-NOS
  - No more explaining what PDD means
Asperger Issue

- Single category: lose Asperger and PDD-NOS
- Scientific validity
- Lack of specificity and sensitivity in separating the diagnoses
- Lack of accurate historical information about very early language development put emphasis on current speech (trainable)
- Overlap in samples when VIQ controlled
- Site specific
Example of Asperger Problem

- Data from Simons Collection
- Over 2200 validated singletons with ASD
- 8500 family members (two biological parents and, in most cases, at least one unaffected sibling) with DNA and intensive behavioral and neuropsychological phenotyping
- Recruited from 12 sites in the US and Canada
- Cell lines and phenotyping data are available through www.sfari.org for interested scientists
ASD Distribution of Probands

Total Probands = 423

<table>
<thead>
<tr>
<th>Site</th>
<th>N</th>
<th>F</th>
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<tbody>
<tr>
<td>aa</td>
<td>32</td>
<td>6.3%</td>
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<tr>
<td>ac</td>
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<tr>
<td>ad</td>
<td>59</td>
<td>11.9%</td>
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<tr>
<td>ae</td>
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<td>ag</td>
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<tr>
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<td>27.3%</td>
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<tr>
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<tr>
<td>aj</td>
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<td>13.3%</td>
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<tr>
<td>ak</td>
<td>24</td>
<td>8.3%</td>
</tr>
<tr>
<td>am</td>
<td>29</td>
<td>17.2%</td>
</tr>
</tbody>
</table>

N = sample size
F = % Females
A = Mean Age
Changes in ASD in the DSM-5

- 3-domain to a 2-domain
  - Communication deficits subsumed under social
How Many Domains?

- Historical
- Population
- Factor Analytic
Some History

- **Kanner had 2**
  - Autistic aloneness
  - Insistence on sameness

- **Wing and Rutter had 3**
  - Social impairment
  - Communication difficulties
  - RRBs
  - Led to DSM criteria

---

In population-based studies

- Past clinical sample in UK
  - Psychiatric and MR registry
  - Evidence for clustering of 3 areas
  - However, clearly those with only 1
  - Social aloofness: more clustering if this was impairment

- Recent studies
  - Problems with selection criteria and Dx
  - Diagnostic variation and heterogeneity
  - Example: 3,000 twin pairs
    - Modest to low correlations between ASD traits in 3 areas
    - This was true even of social and communication (0.2-0.4)

Factor Analytic Approach (ASD)

- One Factor
  - Szatmari et al 2002: ADI-R
  - Constantino et al 2004: SRS; ADI-R
  - Volkmar et al 1988: ABC
  - Wadden et al 1991: ABC

- Three to Six
  - DiLalla and Rogers 1994: CARS
  - Berument et al 1999: ASQ
  - Stella et al 1999: CARS
  - Miranda-Linne and Melin 2002: ABC
  - Tadevosyan-Leyfer et al 2003: ADI-R
  - Lecavalier 2005: GARS
  - Van Lang et al 2006: ADI-R
Factor Analytic Approach

- Mandy and Skuse 2008
  - Reviewed 7 factor studies
  - All studies but one (Constantino 2004) found multiple factors
  - Always social and non-social factors

- Note about RRBs
  - RRB construct may comprise diverse behaviors that do not all share
    the same etiology
  - Three FAs using ADI and ADI-R yielded a two-factor model
    - (Cuccaro et al., 2003; Richler, Bishop, Kleinke, & Lord, 2007; and Szatmari et al., 2006)
      - insistence on sameness
      - repetitive sensory motor actions
  - In two of these, the two factors were independently heritable with
differential relationships to developmental level
DSM-5 Factor Analytic Approach

- Frazier et al., 2012
  - Analyzed symptoms of 14,744 sibs (8,911 ASD, 5,863 non-ASD) from IAN
  - A hybrid model that included both a category (ASD versus non-ASD) and two symptom dimensions (social communication/interaction and restricted/repetitive behaviors) was more parsimonious than all other models and replicated across measures and subsamples.

DSM-5 Factor Analytic Approach

- Mandy, Charman, & Skuse, 2012
  - 708 verbal children, mean age = 9.5 years
  - The DSM-5 model was superior to the three-factor DSM-IV-TR model.
  - It was improved by the removal of items measuring “play and imagination” and “stereotyped and repetitive use of language.”
  - A scale measuring sensory abnormalities was added to the model, and loaded onto its RRB factor.
  - This DSM-5 model fit well in the hold-out sample; was stable across age and sex; and fit adequately in those with clinical and sub-threshold autistic presentations.

Changes in ASD in the DSM-5

- Higher order social impairments

- Relaxing criteria for age of onset

For sub-criterion A.3:

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

DSM-5 recommendations include higher-order impairments in “difficulties adjusting behavior to suit various social contexts.”

For criterion C:

DSM-IV requires that symptoms begin prior to the age of 3 years.

The DSM-5 requires that symptoms begin in early childhood, with the caveat that “symptoms may not become fully manifest until social demands exceed limited capacities; or may be masked by learned strategies in later life.”
Changes in ASD in the DSM-5

- Changes
  - 2 in RRB now
  - By history or current
  - Added sensory
  - New specificity
### 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)
- 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
Autism Spectrum Disorder in DSM-5

Must meet criteria A, B, C, D, & E

A. Persistent deficits in social communication and social interaction across multiple contexts, as manifest by the following, currently or by history (examples illustrative, not exhaustive):

- **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 to social interactions.

- **Deficits in nonverbal 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 total lack of facial expressions and nonverbal communication.

- **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.
Autism Spectrum Disorder in DSM-5

B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least two of the following, currently or by history (examples illustrative, not exhaustive):

- Stereotyped or repetitive motor movements, use of objects, or speech (such as simple motor stereotypies, lining up toys or flipping plates, echolalia, idiosyncratic phrases)

- 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)

- 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).

- 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 smelling or touching of objects, visual fascination with lights or movement)
Autism Spectrum Disorder in DSM-5

C. Symptoms must be present in early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life)

D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning
Autism Spectrum Disorder in DSM-5

E. These disturbances not better explained by intellectual disability (intellectual developmental disorder) or global developmental delays. Intellectual disability and autism spectrum disorder frequently co-occur, to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected for general developmental level.
Autism Spectrum Disorder in DSM-5

• Individuals with a well-established DSM-IV diagnosis of autistic disorder, Asperger’s disorder, or pervasive developmental disorder not otherwise specified should be given the diagnosis of autism spectrum disorder.

• Individuals who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder.
Autism Spectrum Disorder in DSM-5

- Specify:
  - Associated with medical/genetic/environmental
  - Associated with neurodev./mental/bx disorder
  - Severity level
  - With or without intellectual impairment
    - Advise verbal and nonverbal due to unevenness
  - With or without structural language impairment with level of language
  - Catatonia
<table>
<thead>
<tr>
<th>Dimensional Ratings for DSM V ASD</th>
<th>Social Communication</th>
<th>Fixated Interests and Repetitive Behaviors</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Level 3</strong></td>
<td>Severe deficits in verbal and nonverbal. Very limited initiation of social interactions and minimal response to overtures.</td>
<td>Inflexibility of behavior, extreme difficulty coping with change, RRBs that markedly interfere in all spheres. Great distress</td>
</tr>
<tr>
<td>Requires very substantial support</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Level 2</strong></td>
<td>Marked deficits with limited initiations and reduced or atypical responses. Impairment apparent even with supports in place.</td>
<td>Inflexible in behavior, difficulty coping with change, frequent RRBs and interfere in a variety of contexts. Some distress.</td>
</tr>
<tr>
<td>Requires substantial support</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Level 1</strong></td>
<td>With supports in place, noticeable impairments. Difficulty initiating social interactions and clear atypical responses. Maybe decrease social interest.</td>
<td>Behavioral inflexibility causes significant interference in one or more contexts. Trouble switching. Problems organizing and planning.</td>
</tr>
<tr>
<td>Requires support</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Concerns

- Some patients may not get services
- Research data cannot bridge
- Clinicians need retraining
- Patients need to be re-diagnosed to meet criteria
What does the Research Tell Us About Impact?
Studies

• McPartland et al, 2012
  • Used DSM-IV field trial checklist items
  • 933 participants evaluated during the DSM-IV field trial; 657 carried a clinical diagnosis of an ASD, and 276 were diagnosed with a non-autistic disorder
  • Grouped these items into DSM-5 symptoms
  • DSM-5 performed poorly
    • Missed 39%
      • 60.6% of cases with a clinical diagnosis of an ASD met revised DSM-5 diagnostic criteria for ASD. Overall specificity was high, with 94.9%
      • Missed primarily Aspergers and PDD-NOS

Matson et al, 2012

- Two thousand seven hundred and twenty-one toddlers at risk for a developmental disability participated
- DSM-IV and DSM-5 criteria were applied
- DSM-5 resulted in 47.79% fewer toddlers being diagnosed with ASD compared to those on the DSM-IV

But:

- Kappas were applied to test reliability of DSM-5 compared with DSM-IV and all of the “errors” counted against DSM-5 diagnosis.
- Single clinician made both diagnoses – based on retrospective chart review.
- Were DSM-5 data included in original questions?

Studies

- Mattila et al., 2011
  - Used early draft of criteria
  - Epidemiological study of 5,484 8-year-olds in Finland
  - 46% of PDD diagnoses met ASD according to DSM-5
  - DSM-5 draft criteria were shown to be less sensitive to identifying ASDs, particularly those with Asperger's syndrome and some high-functioning subjects with autism
  - When criteria more similar to current DSM-5, went up to 96%
    - Routines and/or rituals vs routines and rituals
    - Included unusual sensory
    - Removed 36 months

Studies

• Frazier et al, 2012
  • Analyzed symptoms of 14,744 sibs (8,911 ASD, 5,863 non-ASD) from IAN
  • Improved specificity over DSM-IV, but worse sensitivity (0.81 vs 0.95)
  • However, from sibs and relaxed criteria

Studies

- Taheri & Perry 2012
  - Chart review of 131 children 2 – 12
  - Diagnoses of AD or PDD-NOS
  - 63% met DSM-V
  - 81% with AD
  - Only 17% of those with PDD-NOS

• Gibbs et al, 2012
  • 132 children
  • The assessment consisted of informal observations, the ADOS, and the ADI-R
  • Of 111 who received DSM-IV-TR diagnosis, 26 did not meet DSM-5
  • 50% of PDD-NOS would not meet DSM-5

Studies

- Swedo et al., 2012
  - The use of datasets and measures designed to address criteria for DSM-IV may not include all the information needed to evaluate properly the DSM-5 criteria which are intended to be broader (so as to include milder symptoms)
  - Studies may differ in terms of using historical versus current ratings
  - The Neurodevelopmental Disorders Workgroup report preliminary data from the DSM-5 field trial suggesting similar rates of ASD in DSM-IV and DSM-5

Studies

• Huerta et al., 2012
  • 4453 kids with PDD and 690 without
  • ADI and ADOS matched to DSM 5 criteria
  • 91% identified
  • As sensitive, with greater specificity with regard to Asperger’s and PDD-NOS

DSM-5 Field Trial

- 2 sites
  - Baystate Medical Center, in Springfield, Mass., and Stanford University, in Palo Alto, Calif
- 63 school aged children, at least 6 years old, English speaking, high risk for ASD
- 2 independent clinicians within 2 weeks
- Reliable across clinicians
- Most who met DSM-IV met DSM-5
  - Those that didn’t, Aspergers, AD, and PDD-NOS

IMFAR 2012 Swedo

- 7500 petitions from “Aspies”
- NY Times article
  - “New Definition of Autism Will Exclude Many”
  - Specifically, 35% of “high-functioning” and “Asperger disorder”
  - Subsequently quoted as “New Criteria Will Deny Services to 65% of Individuals with Autism”
  - DSM panel “backed down”
  - “50% of “Asperger disorder” will be missed, with confirmatory data “in pipeline”
- Wall Street: chill out – not much different
- Will hopefully bring in more diversity (Hispanic, blacks, women)
- Etiologically clear will be removed (e.g., Rett – will use specifier)
IMFAR 2012 Swedo

• Petition:
  • The Asperger’s Association of New England, and individuals who have Asperger Syndrome, their families, and involved professionals
  • 05/04/2012: over 5,000 signed

• Bill in NY:
  • Assembly member Tom Abinanti (D-Greenburgh) of New York introduced Bill, that will define “autism” under New York state law as the currently used criteria set out in the Diagnostic and Statistical Manual-IV-TR (DSM-IV-TR)
IMFAR 2012 Swedo

• Why Asperger out
• New approach will allow:
  • Severity of symptoms
  • Pattern of onset
  • Etiological function
  • Associated conditions
  • Patterns of strengths and weaknesses
IMFAR 2012 Swedo

• Notion of support rather than severity

• Do no Harm:
  • Asperger's must continue to be eligible
  • Considerations of “Aspies” taken into account

• Must be sensitive and specific

• Diagnostic threshold must be sensitive enough to include all ASD, but specific enough to separate “Broader Phenotype” from Autism Spectrum Disorder
New Disorder

• What happens to all the non-RRB individual?
• Social (Pragmatic) Communication Disorder
## Communication Disorders Diagnostic Categories: DSM-5 and DSM-IV

<table>
<thead>
<tr>
<th>DSM-5</th>
<th>DSM-IV</th>
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<tr>
<td>Language Disorder</td>
<td>Expressive Language Disorder</td>
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<tr>
<td>Social (Pragmatic) Communication Disorders</td>
<td>Mixed Receptive-Expressive Language Disorder</td>
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<tr>
<td>Speech Sound Disorder</td>
<td>Phonological Disorder</td>
</tr>
<tr>
<td>Childhood Onset Fluency Disorder</td>
<td>Stuttering</td>
</tr>
<tr>
<td>Unspecified Communication Disorder</td>
<td>Communication Disorder NOS</td>
</tr>
</tbody>
</table>
Social (Pragmatic) Communication Disorder

A. Persistent difficulties in social uses of verbal and nonverbal communication as manifested by all of the following:

1. Deficits in using communication for social purposes, such as greeting and sharing information, in a manner that is appropriate to the social context.

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

3. Difficulties 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. Difficulties understanding what is not explicitly stated (e.g., making inferences) and nonliteral or ambiguous meanings of language (e.g., idioms, humor, metaphors, multiple meanings that depend on the context for interpretation).
Social (Pragmatic) Communication Disorder

B. The deficits result in functional limitations in effective communication, social participation, social relationships, academic achievement, or occupational performance, individually or in combination.
C. The onset of the symptoms is in the early developmental period (but deficits may not become fully manifest until social communication demands exceed limited capacity).

D. The symptoms are not attributable to another medical or neurological condition or to low abilities in the domains of word structure and grammar, and are not better explained by autism spectrum disorder, intellectual disability (intellectual developmental disorder), global developmental delay, or another mental disorder.