Where is the diagnosis of Autism Spectrum Disorders (ASD) going?

Catherine Lord
University of Michigan Autism and Communication Disorders Center (UMACC)
I receive royalties from the publisher of diagnostic instruments but all of my proceeds and those of UMACC collaborators from projects in which we are involved are donated to autism foundations (Have Dreams, Autism Science Foundation).

*Western Psychological Services
Susan Swedo, M.D., pediatrician and chair
Gillian Baird, M.D., developmental pediatrician
Edwin Cook Jr, M.D., child psychiatrist
Francesca Happe, Ph.D., developmental psychologist
James Harris, M.D., child psychiatrist
Water Kaufmann, M.D., neurologist
Bryan King, M.D., child psychiatrist
Catherine Lord, Ph.D., clinical psychologist
Joseph Piven, M.D., child psychiatrist
Sally Rogers, Ph.D., developmental psychologist
Sarah Spence, M.D., child neurologist
Rosemary Tannock, Ph.D., pediatric neuropsychologist
Amy Wetherby, Ph.D., speech–language pathologist
Harry Wright, M.D., child psychiatrist
Outline of talk

- General issues in diagnosis
- Proposed DSM5 autism spectrum criteria
- New Social Communication Disorder diagnosis
- Modifiers and specifiers
- Severity levels of dimensions within ASD
- General comments

- I am not discussing here -- but ask me about -- intellectual disabilities, communication disorders or learning disabilities
A success story for a young man with ASD who has a job and is living independently after a very rough time.
A different profile of another boy at age 9
The same boy at 22 months
What is Autism?

Diagnoses
Where are we now?
Where are we going?
Implications of a diagnosis

Prognosis**
Etiology*
Course**
Appropriate treatments**
Prognosis**
Risk for other difficulties
sk or
Where are we now in diagnosis?

- Worldwide standard criteria (DSM IV/ICD–10)

- With combined history/informant report and direct observation, excellent sensitivity and specificity for prototypic autism in preschool and school age children

- Diagnoses of ASD are generally stable.

- Within a research program, clinical best estimates add to stability of a diagnosis.
Pervasive Developmental Disorders

Autism

- Social Impairment
- Repetitive Behaviors & Restricted Interests
- Speech/Communication Deficits
- Intellectual Disabilities
- Language Disorders
However, the landscape of autism has changed

More referrals of:
  Toddlers and 2 year-olds
  Older children without intellectual disabilities
  Adolescents and adults often with psychiatric comorbidities

Early intervention (and positive effects)

Less association with intellectual disability; children without significant language or cognitive delay present different pictures
Then what?

- Faster diagnoses = narrower comparisons.
- More specific diagnoses = age-related examples.
- Neurobiology = dimensions
1. One spectrum of autistic disorders called Autism Spectrum Disorder (ASD) defined purely by behaviors
   - No differentiation among autism, PDD–NOS, Asperger Syndrome, Childhood Disintegrative Disorder
   - No differentiation within ASD among disorders by etiology (Rett Syndrome, Fragile X, other known genetic disorders)
Many reasons to include Asperger Syndrome & PDD-NOS within one ASD diagnosis

- Scientific validity
  - Questioning the importance of very early language milestones vs. fluent speech in older years
  - Overlap in research when VIQ controlled
- Concern about access to services
Simons Simplex Collection

- Over 2400 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, DNA and phenotyping data are available through www.sfari.org for interested scientists
Simons Simplex Collection

- A publicly available repository of genetic and phenotypic data for well-characterized children with ASD and their families
- Focus is on children likely to have de novo events (in contrast to multiplex families)
- One child with ASD, no known relatives with ASD, at least one sibling and two biological parents without ASD
Simons Simplex Collection
ASD Distribution of Probands

Total Probands = 423

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<th>Site</th>
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N = sample size
F = % Females
A = Mean Age
# Predictors of various ASD diagnoses by site

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Site

Autism
PDD-NOS
Aspergers

N = sample size
F = % Females
A = Mean Age
Care needs to be taken

- That people with diagnoses of Asperger Syndrome or PDD-NOS do not lose services because of being included in ASD
- That people who prefer the term Asperger Syndrome as ways to refer to themselves can use it
- That the ranges of skill levels and abilities within the spectrum of ASD is not underestimated
2. How many domains?

Three existing domains in DSM IV/ICD-10 (social, communication, restricted/repetitive) will become two domains:

Social communication

Restricted interests and repetitive behaviors (RRBs)
Studies within normal populations using brief parent reports often find three moderately correlated factors.
But within samples of children or adults with ASD

- Social–communication skills group are highly correlated and group together with RRBs
- When they do not, differences are primarily accounted for by language level and intelligence
3. For social–communication criteria must be met within EACH subdomain

• *Deficits in social-emotional reciprocity*

• *Deficits in nonverbal communicative behaviors used for social interaction*

• *Deficits in developing and maintaining relationships, appropriate to developmental level*
4. All individuals must have or have had restricted interests and repetitive behaviors (in at least 2 of 4 domains)

A. **Stereotyped or repetitive** speech, motor movements or use of objects

B. Excessive adherence to routines, **ritualized patterns** of verbal or nonverbal behavior or excessive **resistance to change**

C. **Highly restricted, fixated interests** that are abnormal in intensity or focus

D. Hyper- or **hypo-reactivity to sensory input** or unusual interest in sensory aspects of environment
Examples of RRBs
Social Communication Disorder (SCD)

1) is an impairment of pragmatics

2) diagnosed based on difficulty in the social uses of verbal and nonverbal communication in naturalistic contexts,

3) which affects the functional development of social relationships and discourse comprehension and

4) cannot be explained by low abilities in the domains of word structure and grammar or general cognitive ability.
Social Communication Disorder (SCD)

Rule out Autism Spectrum Disorder. Autism spectrum disorder by definition encompasses pragmatic communication problems, but also includes restricted, repetitive patterns of behavior, interests or activities as part of the autism spectrum. Therefore, ASD needs to be ruled out for SCD to be diagnosed.
Social Communication Disorder (SCD)

Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities).
6. Specifiers and modifiers:
With the new criteria, if the child has ASD symptoms, he or she gets an ASD diagnosis with a specifier for the etiology:

ASD with Rett Syndrome
ASD with Fragile X
ASD with 15q11-13

Or a modifier indicating another important factor:

ASD with a language disorder or an intellectual disability
ASD with tonic-clonic seizures
ASD with chronic irritable bowel syndrome
Assessment of overall impairment (for all disorders)

- For a variety of domains relevant to almost any psychiatric condition

- Some of them are:
  - Developmental level or nonverbal and verbal IQ
  - Adaptive functioning
  - Verbal abilities at the time of intake
  - Hyperactivity/impulsivity
  - Sleeping difficulties
  - Co-occurring medical/psychiatric problems or achievement delays
7. Early history is also specified through:

A. Age of perceived onset
B. Pattern of onset (loss? Of what skills?)
C. Examples:
   1) ASD with onset before 18 months and loss of words and social skills
   2) ASD with onset by age 30 months and loss of social skills
   3) ASD with no clear onset and no loss
<table>
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<tr>
<th>Dimensional Ratings for DSM5 ASD</th>
<th>Social Communication</th>
<th>Fixated Interests and Repetitive Behaviors</th>
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<tbody>
<tr>
<td>Requires very substantial support</td>
<td>Minimal social communication</td>
<td>Marked interference in daily life</td>
</tr>
<tr>
<td>Requires substantial support</td>
<td>Marked deficits with limited initiations and reduced or atypical responses</td>
<td>Obvious to the casual observer and occur across context</td>
</tr>
<tr>
<td>Requiring support</td>
<td>Without support, some significant deficits in social communication</td>
<td>Significant interference in at least one context</td>
</tr>
<tr>
<td>Subclinical symptoms</td>
<td>Some symptoms in this or both domains; no significant impairment</td>
<td>Unusual or excessive but no interference</td>
</tr>
<tr>
<td>Normal variation</td>
<td>Maybe awkward or isolated but WNL</td>
<td>WNL for developmental level and no interference</td>
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In the search for biological markers, we need to move to analyses of data from individual children and adults (not just comparisons of averages) with attention to clinically meaningful differences if we want to characterize variation in ASD.
Jalen’s first home-based treatment visit at 19 months
Jalen and mom, after 6 months in the Early Social Interaction program (Wetherby & Wood)
Autism is not all that is problematic for many families and individuals (comorbidities including language delay, intellectual disabilities and other psychological disorders)
In summary

- Can ASD become a disorder like Cerebral Palsy, that implies a constellation of attributes with a clear effect on function?:
  - But has a range of etiologies
  - A range of severities
  - Can be highly impairing or not at all as development progresses
  - Has predictable but different trajectories
  - Is treated or perhaps eventually prevented as we understand its causes
  - Should be addressed in all developmental screenings
The goal is to better understand what goes awry AND WHAT GOES WELL in development in ASD so that we can develop more effective treatments and supports as we search for causes and cures.
DSM V Committee on Neurodevelopmental Disorders

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University of Michigan Autism and Communication Disorders Center (UMACC) Departments of Psychology, Pediatrics and Psychiatry